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This list is an attempt at capturing abstracts for published papers regarding Erdheim-Chester Disease. It is meant for awareness purposes only. It is updated periodically. The last update date appears at the bottom of each page. (Where no PMID is noted, the article was not found on the www.pubmed.gov website.)

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Jul-Aug	Neurol India	Central nervous system Erdheim Chester disease presenting with raised intracranial pressure and cerebellar signs mimicking neurosarcoidosis with secondary cerebral venous thrombosis.	Viswanathan S, Kadir NA, Lip AC, Rafia MH.	Department of Neurology, Kuala Lumpur Hospital, Kuala Lumpur, Malaysia. E-mail: shivenda70@yahoo.com.	No abstract available.	25237960
2014 Sep 17	Rheumatology	Treatment of Erdheim-Chester disease with canakinumab.	Tran TA, Pariente D, Guitton C, Delwail A, Barat-Houari M, Meinzer U.	Assistance Publique Hôpitaux de Paris, Robert Debré University Hospital, University of Paris, Paris, France. tu-anh.tran@chu-nimes.fr.	No abstract available.	25234662
2014 Jul	Indian J Nucl Med	Role of (99m)Tc-MDP bone scan in the diagnosis of Erdheim-Chester disease.	Mukherjee A, Damle N, Bal C, Arora A, Singhal A, Tripathi M, Peepre K.	Dr. Nishikant A Damle, Department of Nuclear Medicine, All India Institute of Medical Sciences, New Delhi - 110 029, Delhi, India. E-mail: nkantdamle@gmail.com	Erdheim-Chester disease (ECD) is a rare systemic non-Langerhans cell histiocytosis. It is a progressive disease of unknown etiology. The (99m)technetium-methylene diphosphonate ((99m)Tc-MDP) bone scan is useful in finding the sites of involvement in the skeleton and is helpful in excluding other causes of bony pain. Also a scintigraphic pattern consistent with ECD should alert the physician to evaluate the patient for visceral sites of involvement using fludeoxyglucose positron emission tomography/computed tomography (FDG PET/CT), as this is known to be fatal at times.	25210284

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Sep 8	Blood	Mutually exclusive recurrent somatic mutations in MAP2K1 and BRAF support a central role for ERK activation in LCH pathogenesis.	Chakraborty R, Hampton OA, Shen X, Simko S, Shih A, Abhyankar H, Lim KP, Covington K, Trevino L, Dewal N, Muzny DM, Doddapaneni H, Hu J, Wang L, Lupo PJ, Hicks MJ, Bonilla DL, Dwyer KC, Berres ML, Poulikakos PI, Merad M, McClain KL, Wheeler DA, Allen CE, Parsons DW.	Texas Children's Cancer Center, Texas Children's Hospital, Houston, TX, United States; ceallen@txch.org.	Langerhans Cell Histiocytosis (LCH) is a myeloproliferative disorder characterized by lesions composed of pathologic CD207+ dendritic cells (DCs) with an inflammatory infiltrate. BRAFV600E remains the only recurrent mutation reported in LCH. In order to evaluate the spectrum of somatic mutations in LCH, whole exome sequencing (WES) was performed on matched LCH and normal tissue samples obtained from 41 patients. Lesions from other histiocytic disorders, juvenile xanthogranuloma (JXG), Erdheim-Chester disease (ECD), and Rosai-Dorfman disease (RDD) were also evaluated. All of the lesions from histiocytic disorders were characterized by an extremely low overall rate of somatic mutations. Notably, 33% (7/21) of LCH cases with wild-type BRAF and none (0/20) with BRAFV600E harbored somatic mutations in MAP2K1 (six in-frame deletions and one missense mutation) that induced ERK phosphorylation in vitro. Single cases of somatic mutations of the MAPK pathway genes ARAF and ERBB3 were also detected. The ability of MAPK pathway inhibitors to suppress MEK and ERK phosphorylation in cell culture and primary tumor models was dependent on the specific LCH mutation. The findings of this study support a model in which ERK activation is a universal endpoint in LCH arising from pathologic activation of upstream signaling proteins.	25202140
2014 Jul	Harefuah	Erdheim-Chester disease	Mazor RD, Shoenfeld Y.	The Zabludowicz Center for Autoimmune Diseases, Sheba Medical Center, Tel Hashomer, Israel. shoenfel@post.tau.ac.il.	Erdheim-Chester disease is an orphan condition which involves the ongoing proliferation, migration and infiltration of the typical CD68(+), CD1a(-) histiocytes to various target foci. Consequently, both the infiltrating and fibrosing elements of the disease promote end organ damage and ultimately, failure. Presentation of the Erdheim-Chester disease typically involves longstanding diabetes insipidus in conjunction with intensifying bone pain that classically affects the femurs and tibiae. Alternatively, the disease may present with neurological deterioration of cerebellar nature. Thus, a high index of clinical suspicion is required when facing a patient with the combination of longstanding diabetes insipidus in conjunction with bone pain or cerebellar dysfunction. Typical symmetric, bilateral increased tracer uptake on a 99mTc bone scintigraphy invoLving the femurs and tibiae, is strongly suggestive of the Erdheim-Chester disease. interferon alpha is considered as the first line of treatment. Nevertheless, recent accumulated data suggests that this disease heavily relies on the Ras/Raf/MEK/ERK signal transduction pathway as inhibition of the V600E mutant BRAF by the small molecule vemurafenib among patients who are carriers of this mutation, yielded dramatic clinical responses.	25189027
2014 Aug 29	Neurology	Marked efficacy of vemurafenib in suprasellar Erdheim-Chester disease	Cohen-Aubart F, Emile JF, Maksud P, Galanaud D, Idbaih A, Chauvet D, Amar Y, Benameur N, Amoura Z, Haroche J.	Groupe Hospitalier Pitié-Salpêtrière, Paris; Université Paris VI Pierre et Marie Curie; E-mail: julien.haroche@psl.aphp.fr.	No abstract available.	25171932

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
Aug 22	Blood	Recurrent RAS and PIK3CA mutations in Erdheim-Chester disease	Emile JF, Diamond EL, Hélias-Rodzewicz Z, Cohen-Aubart F, Charlotte F, Hyman DM, Kim E, Rampal R, Patel M, Ganzel C, Aumann S, Faucher G, Le Gall C, Leroy K, Colombat M, Kahn JE, Trad S, Nizard P, Donadieu J, Taly V, Amoura Z, Abdel-Wahab O, Haroche J.	Human Oncology and Pathogenesis Program and Leukemia Service, Memorial Sloan Kettering Cancer Center, New York, NY, United States; abdelwao@mskcc.org.	Erdheim-Chester Disease (ECD) is a rare histiocytic disorder that is challenging to diagnose and treat. We performed molecular analysis of BRAF in the largest cohort of ECD patients studied to date followed by N/KRAS, PIK3CA, and AKT1 mutational analysis in BRAF wildtype patients. 57.5% (46/80) of patients were BRAFV600E mutant. NRAS mutations were detected in 3/17 ECD BRAFV600E wildtype patients. PIK3CA mutations (p.E542K, p.E545K, p.A1046T and p.H1047R) were detected in 7/55 patients, of whom 4 also had BRAF mutations. Mutant NRAS was present in peripheral blood CD14+ cells, but not lymphoid cells, from an NRASQ61R mutant patient. Our results underscore the central role of RAS-RAF-MEK-ERK activation in ECD and identify an important role of activation of RAS-PI3K-AKT signalling in ECD. These results provide a rationale for targeting mutant RAS or PI3K/AKT/mTOR signalling in the subset of ECD patients with NRAS or PIK3CA mutations.	25150293
2014 Aug 19	Tidsskr Nor Laegeforen	A man in his forties with swelling in both orbits].	Midtvedt O, Gran JT, Solheim H, Kirkhus E, Spetalen S.	Revmatologisk avdeling Oslo universitetssykehus, Rikshospitalet.	ERDHEIM-CHESTER DiseaseA multi-disiplinary challengeThe histiocytoses are a diverse, but rare group of disorders with symptoms affecting many organs, varying from self-limiting, localised lesions to disseminated multi-organ disease. The diagnostic challenges are illustrated and discussed in the following case.Case report. A man in his forties was admitted to hospital due to pain in his right eye and visual disturbances. MRI imaging detected a mass in his right orbit and a minor mass in his left orbit. The histological results of the mass in his right orbit revealed an inflammatory process with lymphocytes and macrophages and no sign of vasculitis, infection or malignancy. The diagnosis pseudotumor orbita was made and treatment with corticosteroids was initiated. He did not respond to corticosteroids or radiotherapy and increasing symptoms necessitated rehospitalisation. Further tests disclosed a multisystem disease which affected the aorta, skeleton, lung, heart and kidney. The biopsy was reconsidered and the disease was classified as a histiocytosis with CD68 positive and CD1a negative cells. The diagnosis Erdheim-Chester was given, about 14 months after the initial hospitalisation. Treatment with interferon α was started.	25138406

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014	Klin Onkol	Impact of anakinra treatment on cytokine and lymphocytes/monocytes profile of an Erdheim-Chester patient.	Sevčíková S, Kubiczková L, Sedlaříková L, Říhová L, Kryukov F, Szturz P, Hajek R, Pour L, Adam Z.	RNDr. Sabina Ševčíková, Ph.D.; Babak Myeloma Group; Department of Pathological Physiology; Faculty of Medicine; Masaryk University; Kamenice 5, A4; 625 00 Brno; Czech Republic; e-mail: sevcik@med.muni.cz	Abstract BACKGROUND: Erdheim-Chester disease (ECD) is a rare non-Langerhans cells histiocytosis associated with intense immune activation. In our clinical center, an ECD patient was treated with anakinra, IL1RA (interleukin1 receptor antagonist), resulting in clinical improvement and major decrease of pathological fatigue. The aim of the study was to evaluate changes in cytokine profile and shift of immune cells estimated by flow cytometric analysis of ECD patient before, during initial stages of anakinra treatment as well as after treatment ceased in comparison to healthy donors. METHODS: Singleplex reactions of 19 individual cytokines from serum of ECD patient were measured by FACS array. Flow cytometric analyses were performed on peripheral blood cells. RESULTS: The most striking result is substantial decrease of IL6 immediately after anakinra treatment started suggesting a major role of IL1 pathway in ECD pathophysiology. As for flow cytometric analysis, increased number of CD16+ monocytes before treatment is a new finding. CONCLUSION: Our results suggest that IL6 may be a marker of early treatment response of ECD patients treated with anakinra.	25115717
2014 May-Jun	Vnitr Lek	Erdheim-Chester disease-the rare and under-diagnosed disease-editorial	Mistrík M.	Laboratory of Genome Integrity; Institute of Molecular and Translational Medicine; Palacky University; Olomouc, Czech Republic. martin.mistrik@upol.cz	[PET-CT documented complete remission of Erdheim-Chester disease, lasting more than 4 years from treatment initiation with cladribine. [Article in Czech]	25069140
2014 Jul	Arq Neuropsiquiatr	Histiocytosis: a review focusing on neuroimaging findings.	Gabbay LB, Leite Cda C, Andriola RS, Pinho Pda C, Lucato LT.	Larissa Barcessat Gabbay; Av. Dr. Enéas de Carvalho Aguiar, s/n° / Rua 1 Cerqueira César; 05403-900 São Paulo SP, Brasil; E-mail: larissagabbay@hotmail.com	OBJECTIVE: Histiocytosis is a systemic disease that usually affects the central nervous system. The aim of this study is to discuss the neuroimaging characteristics of Langerhans cell histiocytosis (LCH), the most common of these diseases; and the non-Langerhans cells histiocytosis (NLCH), which includes entities such as hemophagocytic syndrome, Erdheim-Chester and Rosai-Dorfman diseases. METHOD: Literature review and illustrative cases with pathologic confirmation. RESULTS: In LCH, the most common findings are 1) osseous lesions in the craniofacial bones and/or skull base; 2) intracranial, extra-axial changes; 3) intra-axial parenchymal changes (white and gray matter); 4) atrophy. Among the NLCH, diagnosis usually requires correlation with clinical and laboratory criteria. The spectrum of presentation includes intraparenchymal involvement, meningeal lesions, orbits and paranasal sinus involvement. CONCLUSION: It is important the recognition of the most common imaging patterns, in order to include LCH and NLCH in the differential diagnosis, whenever pertinent.	25054989

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Jun 16	Case Rep Hematol	High-dose methotrexate for the treatment of relapsed central nervous system erdheim-chester disease.	Ho P, Smith C.	Department of Clinical Haematology, Austin Health, Heidelberg, VIC, Australia. Email: prahladho@gmail.com	Erdheim-Chester disease (ECD) is a rare multisystem non-Langerhans histiocytosis. CNS involvement is a major complication, which is often rapidly progressive and confers a poor prognosis. However, treatment of CNS ECD is difficult due to poor CNS penetrance by the most effective chemotherapeutic drugs commonly used in this disorder (e.g., interferon and cladribine). We describe a case of a 60-year-old lady with a 5-year history of stable systemic ECD who presented with new brainstem lesions and rapid, steroid-refractory neurological deterioration which required immediate intervention. High-dose methotrexate was chosen due to its rapid onset of action and excellent CNS penetration. Her neurological deterioration was quickly arrested with significant functional improvement, which was sustained for 4 months with consolidation doses of high-dose methotrexate. Subsequent treatment with cladribine and interferon did not confer any appreciable clinical improvement. High-dose methotrexate is effective in controlling rapidly progressive CNS ECD and should be considered as a salvage agent prior to commencement of more definitive treatment.	25031876
2014 Jul 9.	Eur J Intern Med	An enlightening scan.	Canziani L, Tomelleri A, Cavalli G.	Vita-Salute San Raffaele University, Italy. Electronic address: l.canziani@studenti.unisr.it.	No abstract available.	25023920
2014 Aug	Insights Imaging	Thoracic, abdominal and musculoskeletal involvement in Erdheim-Chester disease: CT, MR and PET imaging findings.	Antunes C, Graça B, Donato P.	Centro Hospitalar e Universitário de Coimbra, Praceta Prof. Mota Pinto, 3000-075, Coimbra, Portugal, celita_msa@hotmail.com.	BACKGROUND: Erdheim-Chester disease (ECD) is a rare, non-Langerhans cell histiocytosis with characteristic radiological and histological features. This entity is defined by a mononuclear infiltrate consisting of lipid-laden, foamy histiocytes that stain positively for CD68 and negatively for CD1a. Osseous involvement is constant and characteristic. Extra-osseous lesions may affect the retroperitoneum, lungs, skin, heart, brain and orbits. METHODS: Both radiography and technetium-99m bone scintigraphy may reveal osteosclerosis of the long bones, which is a typical finding in ECD. For visceral involvement, computed tomography (CT) is most useful, while magnetic resonance (MR) imaging is more sensitive for cardiovascular lesions; 2-[fluorine-18] fluoro-2-deoxy-d-glucose (FDG) positron emission tomography (PET)/CT scanning is useful in assessing the extension of ECD lesions. RESULTS: The prognosis is extremely variable and is often worse when there is cardiovascular system involvement. Diagnosis is based on the combination of radiographic, CT, MR imaging and nuclear medicine features and a nearly pathognomonic immunohistochemical profile. CONCLUSION: The aims of this work are to perform a systematic review of Erdheim-Chester disease as seen on imaging of the chest, abdomen and musculoskeletal system and to discuss the diagnostic workup and differential diagnoses according to the imaging presentation. Teaching points • Bone involvement is usually present in patients, and the imaging findings are pathognomonic of ECD. • The circumferential periaortic infiltration may extend to its branches, sometimes becoming symptomatic. • Cardiac involvement-the pericardium, right atrium and auriculoventricular sulcus-worsens its prognosis. • Perirenal infiltration extending to the proximal ureter is highly suggestive of this disease.	25017251

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Oct	Cornea	Bilateral adult epibulbar xanthogranulomas suspicious for erdheim-chester disease.	Aggarwal S, Jakobiec FA, Hamrah P.	Cornea & Refractive Surgery Service, Massachusetts Eye and Ear Infirmary, Department of Ophthalmology, Harvard Medical School, Boston, MA	PURPOSE: The aim of this study was to report the clinical, imaging, and histopathological findings of bilateral, conjunctival adult-onset xanthogranulomas that raised the prospect of a mild form of Erdheim-Chester disease. METHODS: This is a case report. RESULTS: A 35-year-old white male complaining of ocular irritation, presented with bilateral, nasal and temporal, yellow, elevated conjunctival lumps first noticed 1.5 years back, which were not associated with other ocular findings. The lesions were firm, attached to the underlying episclera, and measured 1.1 × 0.9, 1.1 × 0.8, 1.2 × 0.5, and 0.5 × 0.5 cm in the temporal and nasal right and left eyes, respectively. Each mass was fleshy with vascularity at the peripheral margin. Histopathologic evaluation after excisional biopsy revealed lipidized xanthoma cells, multiple Touton giant cells, and lymphocytes. Immunohistochemical staining was positive for adipophilin (lipid), CD68, CD163 histiocytes, CD3 T cells (with CD8 cytotoxic T cells > CD4 T-helper cells), and virtually no CD20 B cells or IgG4 plasma cells. The patient later acquired similar xanthogranulomatous subcutaneous lesions on the extremities. Positron emission tomography scans showed sclerosis in the medullary cavities of the tibia and the radius of both legs and arms, and an absence of retroperitoneal lesions. A normal serum immunoelectrophoresis and the absence of a BRAF gene mutation were demonstrated. CONCLUSIONS: Adult-onset xanthogranuloma can present as a solitary conjunctival mass without periocular or orbital involvement. The clinical, histopathologic, and radiologic findings in this case are suggestive of Erdheim-Chester disease without displaying any life-threatening lesions to date. Histopathologic and imaging studies can help in obtaining a diagnosis. Ophthalmologists should be aware that xanthogranulomatous conditions may have potential systemic implications, and a thorough systemic evaluation is recommended for lesions that initially seemed to be isolated in nature.	25014153
2014 Jul 7	World J Gastroenterol	Erdheim Chester - a rare disease with unique endoscopic features.	Ben-Yaakov G, Munteanu D, Sztarkier I, Fich A, Schwartz D.	Gil Ben-yaakov, MD, Institute of Gastroenterology and Hepatology, Soroka University Medical Center, Faculty of Health Science Ben Gurion University of the Negev, Beer sheva 84101, Israel. Email: gilbenyakov@gmail.com	Erdheim-Chester disease (ECD) is a rare inflammatory syndrome in which systemic infiltration of non-Langerhans cell histiocytes occurs in different sites. Both the etiology and pathophysiology of ECD are unknown, but CD68 positive CD 1a/S100 negative cells are characteristic. The presentation of ECD differs according to the involved organs. This case report describes a patient with ECD and the gastrointestinal manifestations and unique endoscopic appearance as seen in gastroscopy and colonoscopy with histological proof of histiocyte infiltration of the lamina propria. The clinical and endoscopic findings of this unique case, to our knowledge, were never described before, so were the features of the gastrointestinal involvement in this disease.	25009409

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Jun 15	Oncotarget	BRAF V600E mutations in urine and plasma cell-free DNA from patients with Erdheim-Chester disease.	Janku F, Vibat CR, Kosco K, Holley VR, Cabrilo G, Meric-Bernstam F, Stepanek VM, Lin PP, Leppin L, Hassaine L, Poole JC, Kurzrock R, Erlander MG.	Department of Investigational Cancer Therapeutics, The University of Texas MD Anderson Cancer Center. Email: fjanku@mdanderson.org	Erdheim-Chester disease (ECD) is a rare histiocytosis with a high prevalence of BRAF V600E mutation (>50% of patients). Patients with BRAF-mutant ECD can respond to BRAF inhibitors. Unfortunately, the lack of adequate archival tissue often precludes BRAF testing. We hypothesized that cell-free DNA (cfDNA) from plasma or urine can offer an alternative source of biologic material for testing. We tested for BRAF V600E mutation in cfDNA from the plasma and urine of 6 ECD patients. In patients with available archival tissue, the result of BRAF mutation analysis was concordant with plasma and urine cfDNA results in all 3 patients (100% agreement, kappa 1.00). In all 6 patients, BRAF mutation analysis of plasma and urine cfDNA was concordant in 5 of 6 patients (83% agreement, kappa 0.67). Testing for BRAF V600E mutation in plasma and urine cfDNA should be further investigated as an alternative to archival tissue mutation analysis.	25003820
2014 Jul 2	Med Clin (Barc).	Erdheim-Chester disease	García-Gómez FJ, Acevedo-Báñez I, Rivas-Infante E, Borrego-Dorado I.	1Servicio de Medicina Nuclear, Hospital Universitario Virgen del Rocío, Sevilla, España. Electronic address: javier191185@gmail.com.	No abstract available. [Article in Spanish]	24998099
2014 Jun 13	Front Immunol.	Oncogene-induced senescence as a new mechanism of disease: the paradigm of erdheim-chester disease	Cavalli G, Biavasco R, Borgiani B, Dagna L.	Lorenzo Dagna, Unit of Internal Medicine and Clinical Immunology, Vita-Salute San Raffaele University, IRCCS San Raffaele, Via Olgettina 60, Milano 20132, Italy e-mail: Lorenzo.Dagna@unisr.it	Erdheim-Chester disease (ECD) is a rare form of systemic histiocytosis characterized by the diffuse infiltration of tissues by lipid-laden macrophages. As the clinical course and prognosis are highly influenced by site of disease involvement, ECD course ranges from asymptomatic to life threatening, with a reported global 5-year mortality of 30-40%. Whether ECD is an inflammatory or clonal disease in its nature has long been debated. The disease is characterized by a network of pro-inflammatory cyto/chemokines responsible for the recruitment and activation of histiocytes into ECD lesions, similarly to what reported in Langerhans cell histiocytosis (LCH). Growing evidence supports a central role of the oncogenic BRAF(V600E) mutation in histiocytosis pathogenesis, and suggests oncogene-induced senescence (OIS), a major protective mechanism against oncogenic events characterized by cell-cycle arrest and the induction of pro-inflammatory molecules, as the possible link between the oncogenic mutation and the observed inflammation. Indeed, ECD recapitulates in vivo the molecular events associated with OIS, i.e., cell-cycle arrest and a potent local inflammatory response. Accordingly, the infiltration of different tissues by macrophages and the inflammatory local and systemic effects observed in ECD likely represent a drawback of OIS. Therefore, these findings delineate a new conception of OIS as a new pathogenic mechanism intrinsically responsible for disease development.	24982657
2014 Jul	Anticancer Res.	Erdheim-Chester disease: a comprehensive review.	Abdelfattah AM, Arnaout K, Tabbara IA.	Division of Hematology and Oncology, Department of Medicine, George Washington University Medical Center, Washington, DC, U.S.A. itabbara@mfa.gwu.edu.	Erdheim-Chester disease is a rare form of non-Langerhans' cell histiocytosis characterized by multi-system infiltration by xanthogranulomas composed of foamy histiocytes surrounded by fibrosis. Approximately 400 cases have been reported in the literature, and the recent increase in the number of cases is likely due to the increased awareness of its associated morbidity and mortality. The etiology of this disease remains unknown, the clinical course is variable and treatment is still not well-established. The objective of this review is to describe the pathogenesis, clinical manifestations, and diagnosis of this rare disorder, and to review its prognosis and treatment. Erdheim-Chester disease (ECD) is a rare form of non-Langerhans' cell histiocytosis. It was first described in 1930. Approximately 400 cases have been reported in the literature.	24982329

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 May-Jun	Vnitr Lek.	PET-CT documented complete remission of Erdheim-Chester disease, lasting more than 4 years from treatment initiation with cladribine].	Adam Z, Řehák Z, Koukalová R, Bortlíček Z, Krejčí M, Pour L, Szturz P, Prášek J, Nebeský T, Adamová Z, Král Z, Mayer J.	Interní hematoonkologická klinika, LF MU a FN Brno. z.adam@fnbrno.cz	Erdheim-Chester disease is a very rare histiocytic disease. It represents one form of juvenile xanthogranuloma in WHO classification of blood diseases. The disease often causes B symptoms, skeletal pain and also may cause diabetes insipidus and retroperitoneal fibrosis. Selection of therapy depends on published case reports and small clinical trials. There are no recommendations for treatment based on randomized studies. Interferon α is probably the most commonly used drug for this disease. Some remissions have been described after treatment. However, long-term interferon α application is needed which is associated with numerous side effects. There are limited experiences with clabridine in this indication. In Pubmed Medline database, we have found 3 publications dedicated to description of treatment response after cladribine in Erdheim-Chester disease and other 7 papers evaluating effect of cladribine on juvenile xanthogranuloma forms, mostly with positive outcome. Based on these 10 publications we choose cladribine as first-line treatment in our patient. The treatment started in October 2009 with combination of 2-chlorodeoxyadenosine (Litak) 5 mg/m2 sc. + cyclophosphamide 150 mg/m2 iv. + dexamethasone 24 mg iv., five days consecutively. These cycles were repeated monthly. Mentioned formula was submitted 4 times and 3 times in limited application on day 1 - 3. The reason of that was neutropenia grade 3. All symptoms disappeared after treatment. Only diabetes insipidus persisted because damage of pituitary stalk is irreversible. Therapeutic effect was monitored by PET-CT imaging, initially every 6 months, later in 12-month intervals. PET-CT imaging showed complete remission of disease and 4.5 years duration of remission after treatment. The treatment was well tolerated with no complications implying hospitalization. Only mild thrombocytopenia and neutropenia remains after 4.5 years. Based on case report and publications we consider cladribine as appropriate firs-line drug for Erdheim-Chester disease. Therapeutic failure after 3-4 cycles may suggest other options (interferon α, anakinra, vemurafenib), but only in the case if healthcare provider is willing to cover this new and more expansive treatment than therapy with cladribine.	24974755
2014 Jun 1	Tex Heart Inst J.	Multidetector computed tomographic imaging of Erdheim-Chester disease.	Yuceler Z, Kantarci M, Karabulut N, Ogul H, Bayraktutan U, Akman C.	Mecit Kantarci, MD, 200 Evler Mah. 14. Sok No:5, Dadaskent, 25090 Erzurum, Turkey moc.liamtoh@darnakka	Erdheim-Chester disease is a rarely reported disease that can affect nearly every organ and chiefly infiltrates the connective, perivascular, and adipose tissue. The disease is a form of non-Langerhans-cell histiocytosis characterized by the proliferation of foamy histiocytes; its cardiovascular complications carry a severe prognosis. We present the case of a 29-year-old woman who was admitted for analysis of her angina. Our evaluation with use of cardiac multidetector computed tomographic angiography revealed large mediastinal soft tissue that compressed the patient's left anterior descending coronary artery. To our knowledge, this is the first report of the use of low-dose, dual-source, 256-slice multidetector computed tomography to characterize Erdheim-Chester disease that exclusively caused angina and stenosis of a coronary artery in a young adult.	24955059
2014 May	Ir Med J	Gastrointestinal Erdheim-Chester disease.	Tevlin R, Cahalane AM, Larkin JO, Treacy A, Connaghan D, Winter DC.	Centre for Colorectal Disease, St. Vincent's University Hospital, Dublin 4, Ireland. Electronic address: Des.winter@gmail.com.	We report a rare case of Erdheim-Chester Disease, a non-Langerhans cell histiocytosis. A 60-year old female presented with a seven-month history of vague abdominal symptoms. A large retroperitoneal mass was detected on computed tomography (CT), but multiple CT-guided biopsy samples were inconclusive. Laparoscopy revealed a mass in the distal ileum, which was resected. Histology and immuno-histochemistry supported a diagnosis of Erdheim-Chester Disease.	24908861

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Aug 14	Blood	Association of both Langerhans cell histiocytosis and Erdheim-Chester disease linked to the BRAFV600E mutation	Hervier B, Haroche J, Arnaud L, Charlotte F, Donadieu J, Néel A, Lifermann F, Villabona C, Graffin B, Hermine O, Rigolet A, Roubille C, Hachulla E, Carmoi T, Bézier M, Meignin V, Conrad M, Marie L, Kostrzewa E, Michot JM, Barete S, Taly V, Cury K, Emile JF, Amoura Z; French Histiocytoses Study Group.	Service de médecine interne 2, Centre national de référence maladies systémiques rares, groupe hospitalier Pitié-Salpêtrière, AP-HP, UPMC, université Paris-6, 47-83, boulevard de l'Hôpital, 75651 Paris, France. Electronic address: julien.haroche@psl.aphp.fr	Histiocytoses are a group of heterogeneous diseases that mostly comprise Langerhans cell histiocytosis (LCH) and non-LCH. The association of LCH with non-LCH is exceptional. We report 23 patients with biopsy-proven LCH associated with Erdheim-Chester disease (ECD) (mixed histiocytosis) and discuss the significance of this association. We compare the clinical phenotypes of these patients with those of 56 patients with isolated LCH and 53 patients with isolated ECD. The average age at diagnosis was 43 years. ECD followed (n = 12) or was diagnosed simultaneously with (n = 11) but never preceded LCH. Although heterogeneous, the phenotype of patients with mixed histiocytosis was closer to that of isolated ECD than to that of isolated LCH (principal component analysis). LCH and ECD improved in response to interferon alpha-2a treatment in only 50% of patients (8 of 16). We found the BRAF(V600E) mutation in 11 (69%) of 16 LCH lesions and in 9 (82%) of 11 ECD lesions. Eight patients had mutations in both ECD and LCH biopsies. Our findings indicate that the association of LCH and ECD is not fortuitous and suggest a link between these diseases involving the BRAF(V600E) mutation.	24894769
2014 May 27	Rev Med Interne.	Erdheim-Chester disease	Haroche J, Cohen-Aubart F, Arnaud L, Hervier B, Charlotte F, Drier A, Gorochov G, Grenier PA, Cluzel P, Maksud P, Emile JF, Amoura Z.	Service de médecine interne 2, Centre national de référence maladies systémiques rares, groupe hospitalier Pitié-Salpêtrière, AP-HP, UPMC, université Paris-6, 47-83, boulevard de l'Hôpital, 75651 Paris, France. Electronic address: julien.haroche@psl.aphp.fr	Erdheim-Chester disease is a rare and orphan disease. Despite having been overlooked previously, numerous new cases have been diagnosed more recently. The number of Erdheim-Chester disease cases reported has increased substantially: more than 300 new cases have been published in the past 10 years. This situation is mainly a result of the generally better awareness among pathologists, radiologists, and clinicians of various aspects of this rare disease. The field has been particularly active in the last few years, with evidence of the efficacy of interferon-α, the description of a systemic pro-inflammatory cytokine signature, and most recently, reports of the dramatic efficacy of BRAF inhibition in severe, BRAFV600E mutation-associated cases of Erdheim-Chester disease. Also, BRAF mutations have been found in more than half of the patients with Erdheim-Chester disease who were tested. Detailed elucidation of the pathogenesis of the disease is likely to lead to the development of better targeted and more effective therapies.	24878295
2014 May 28	Clin Nucl Med.	Visualization of Orbital Involvement of Erdheim-Chester Disease on PET/CT.	Beylergil V, Carrasquillo JA, Hyman DM, Diamond EL.	Molecular and Imaging Therapy Service, Department of Radiology, Memorial Sloan-Kettering Cancer Center, NY, NY	We report a 58-year-old man who presented with swelling and redness in his left eye, headache, and blurred vision. A contrast-enhanced CT of the orbits revealed bilateral orbital masses. Whole-body PET/CT showed bilateral retrobulbar hypermetabolic soft tissue lesions, multiple areas of soft tissue involvement, and osseous lesions in bilateral lower extremities. An open surgical biopsy of the left orbital mass revealed xanthomatous non-Langerhans histiocytic infiltrate with Touton giant cells, positive for CD68 but negative for CD1a, establishing a diagnosis of Erdheim-Chester disease.	24873784

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014	Am Soc Clin Oncol Educ Book	Biological and Therapeutic Implications of the BRAF Pathway in Histiocytic Disorders	Arceci RJ	Children's Center for Cancer and Blood Disorders, Hematology/Oncology, Ron Matricaria Institute of Molecular Medicine, Phoenix Children's Hospital, University of Arizona, College of Medicine, Phoenix, AZ	Langerhans cell histiocytosis (LCH) has historically evolved in its classification from a primary immune dysregulatory disorder to what current evidence supports as a dendritic cell neoplasm with an immune-inflammatory component. A key part of the classification of LCH as a neoplasm has been the identification of BRAF V600E mutations in 35% to 60% of cases. Tumor protein p53 (TP53) and RAS mutations have also been identified, albeit in less than 2% of reported cases. Of note, over 50% of patients with another dendritic cell disease, Erdheim-Chester Disease, have also been shown to have BRAF V600E mutations. Although the BRAF mutations have not been shown to be associated with extent of disease, they may still provide a target for a molecularly guided approach to therapy. In cases of LCH in which no BRAF mutations were identified, there was evidence for activation of the RAS-RAF-MEK-extracellular signal-regulated kinases (ERK) pathway, suggesting that similar to other tumors, this pathway may be therapeutically exploitable. Anecdotal responses have been reported in a few patients with LCH and Erdheim-Chester Disease to vemurafenib, a BRAF V600E inhibitor. Although these results pave the way for careful, prospective clinical testing, selection of the optimal groups in which to test such inhibitors, alone or in combination, will be critical based on the toxicity profile thus far observed in adults with melanoma and other BRAF mutated tumors.	24857137
2014 May 21	Blood	Consensus guidelines for the diagnosis and clinical management of Erdheim-Chester disease (available in English and Spanish – email support@erdheim-chester.org)	Diamond EL, Dagna L, Hyman DM, Cavalli G, Janku F, Estrada-Veras J, Ferrarini M, Abdel-Wahab O, Heaney ML, Scheel PJ, Feeley NK, Ferrero E, McClain KL, Vaglio A, Colby T, Arnaud L, Haroche J.	Department of Neurology, Memorial Sloan-Kettering Cancer Center, New York, NY, United States; diamone1@mskcc.org	Erdheim-Chester Disease (ECD) is a rare non-Langerhans histiocytosis. Recent findings suggest that ECD is a clonal disorder, marked by recurrent BRAFV600E mutations in more than 50% of patients, in which chronic uncontrolled inflammation is an important mediator of disease pathogenesis. Although approximately 500-550 cases have been described in the literature to date, increased physician awareness has driven a dramatic increase in ECD diagnoses over the last decade. ECD frequently involves multiple organ systems and has historically lacked effective therapies. Given the protean clinical manifestations and the lack of a consensus-derived approach for the management of ECD, we provide here the first multidisciplinary consensus guidelines for the clinical management of ECD. These recommendations were outlined at the First International Medical Symposium for ECD, comprised of a comprehensive group of international academicians with expertise in the pathophysiology and therapy of ECD. Detailed recommendations on the initial clinical, laboratory, and radiographic assessment of ECD patients are presented in addition to treatment recommendations based on critical appraisal of the literature and clinical experience. These formalized consensus descriptions will hopefully facilitate ongoing and future research efforts in this disorder.	24850756

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 May 7	Mayo Clin Proc.	Erdheim-Chester Disease: Characteristics and Management	Munoz J, Janku F, Cohen PR, Kurzrock R	Hematology-Oncology, Banner MD Anderson Cancer Center, Gilbert, AZ. E-mail: javier.munoz@me.com	Erdheim-Chester disease is a rare CD68+, CD1a- non-Langerhans cell histiocytosis with multiorgan involvement. The etiology of Erdheim-Chester disease is unclear; there are no known associated infectious or hereditary genetic abnormalities. However, somatic BRAF mutations have recently been identified in these patients. Historically, the literature regarding the management of Erdheim-Chester disease consisted of case reports and small case series with anecdotal therapeutic responses to agents including, but not limited to, cytotoxic chemotherapy, bone marrow transplantation, cladribine, corticosteroids, IFN-α, the BCR-ABL/KIT inhibitor imatinib mesylate, the IL-1 receptor antagonist anakinra, the TNF-inhibitor infliximab, and recently the BRAF inhibitor vemurafenib. We performed a search of the literature using PubMed with the terms Erdheim Chester disease, without date limitations, including case reports, case series, original articles, and previous review articles. In the absence of large-scale studies, experience-based management prevails. The present review details our approach to the management of patients with Erdheim-Chester disease.	24814521
2014 Apr;30	J Anaesthesiol Clin Pharmacol.	Erdheim - Chester disease: Clinical pearls for the anesthesiologist	Hariharan U, Goel AV, Sharma D.	Dr. Uma Hariharan, Department of Onco-Anaesthesia, Rajiv Gandhi Cancer Institute and Research Center, Rohini, New Delhi - 110 085, India. E-mail:	n/a	24803787
2014 Apr 27	Urology	Urologic Manifestations of Erdheim-Chester Disease	Yelfimov DA, Lightner DJ, Tollefson MK.	Department of Urology, Mayo Medical School and Mayo Clinic, Rochester, MN. E-mail: tollefson.matthew@mayo.edu	OBJECTIVE: To describe the urologic manifestations of Erdheim-Chester disease (ECD). ECD is a rare multisystem disorder of non-Langerhans cell histiocytosis. In addition to classic long bone involvement, the retroperitoneum is a well-established site of disease infiltration. Herein, we present the urologic manifestations and outcomes of ECD in a large series of patients. METHODS: We identified 47 patients diagnosed with ECD between 1996 and 2012 at our institution. The medical records and imaging for these patients were reviewed for urologic involvement, including perirenal soft tissue encasement, renal atrophy, hydronephrosis, chronic renal insufficiency, diabetes insipidus, and lower urinary tract symptoms. RESULTS: At diagnosis, the median patient age was 57 years (interquartile range 49, 68), and median follow-up after diagnosis was 3 years (interquartile range 1.8, 7.3). There were 31 male patients (66%) and 16 female patients (34%). The majority of these patients (37, 79%) demonstrated evidence of urologic involvement from ECD, requiring urologic surgery in 13 (28%). This urologic involvement included retroperitoneal infiltration (28, 60%), worsening lower tract urinary symptoms from diabetes insipidus (21, 45%), hydronephrosis (10, 21%), and chronic kidney disease (18, 38%). CONCLUSION: The incidence of urologic involvement with ECD is higher than previously reported in the published data. Urologists should be aware of this disorder, as it might mimic other retroperitoneal diseases and might contribute to lower urinary tract symptoms, hydronephrosis, renal atrophy, and chronic kidney disease.	24788188

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Jun 15	Int J Cardiol.	Constrictive pericarditis in Erdheim-Chester disease: An integrated echocardiographic and magnetic resonance approach	Palazzuoli A, Mazzei MA, Ruocco G, Volterrani L	Department of Medical, Surgical and Neuro Sciences, Internal Medicine Unit Cardiology Section, Le Scotte Hospital, Siena, Italy. E-mail: palazzuoli2@unisi.it	Erdheim–Chester disease (ECD) is a rare non-Langerhans histiocytosis affecting middle-aged adults. The disease is named after the cardiology fellow (William Chester) and his mentor (Jakob Erdheim) who reported the first two cases in 1930 in Vienna as 'lipoid granulomatosis'. Its etiology is unknown, and it is characterized by the proliferation of lipid-containing foamy histiocytes infiltrating bones and potentially every organs. Histiocytic infiltration leads to xanthogranulomatous infiltrates of multiple organ systems/	24768396
2014 Apr 23	Histopathology.	An NRAS mutation in a case of Erdheim Chester disease	Aitken SJ, Presneau N, Tirabosco R, Amary MF, O'Donnell P, Flanagan AM.	Research Department of Pathology, UCL Cancer Institute, London, UK; Sarah Cannon/University College London Advanced Diagnostics Molecular Profiling Research Laboratories, London, UK	Erdheim Chester disease (ECD) is a rare, non-Langerhans' cell histiocytic neoplastic disorder that usually presents in patients in their fifties. ECD often presents as a multi-system macrophagic infiltration and common sites of involvement are the central nervous system, cardiovascular system, respiratory system, retroperitoneum, and skin. Extra-skeletal involvement is usually responsible for death due to secondary complications and ECD has had until recently an extremely poor prognosis (vide infra), with less than half of patients surviving three years.	24754681
2014 Apr 23	Mod Rheumatol.	Rapid progression to cardiac tamponade in Erdheim-Chester disease despite treatment with interferon alpha.	Nakhleh A, Slobodin G, Elias N, Bejar J, Odeh M.	Prof. Majed Odeh, Department of Pathology, Bnai Zion Medical Center, Haifa, Israel. Tel: 972-4-8358781. Fax: 972-4-8359790. E-mail: majed.odeh@b-zion.org.il	Erdheim-Chester disease (ECD) is a rare form of non-Langerhans histiocytosis with heterogeneous clinical manifestations. The most common presentation is bone pains typically involving the long bones. Approximately 75% of the patients develop extraskeletal involvement. Cardiac involvement is seen in up to 45% of the patients, and although, pericardial involvement is the most common cardiac pathology of this rare disease, cardiac tamponade due to ECD has been very rarely reported. We describe a case of a patient found to have ECD with multi-organ involvement and small pericardial effusion, which progressed to cardiac tamponade despite treatment with interferon alpha.	24754271
2014 Apr 16	J Neuroophthalmol.	Homonymous Hemianopia Due to Erdheim-Chester Disease	Hills WL, Nassef AH, Grafe MR, Weissman JL, Moster SJ, Falardeau J, Mardekian SK, Curtis MT, Moster ML.	Department of Neuro-Ophthalmology (WLH, JF), Casey Eye Institute, Oregon Health & Science University, Portland, Oregon	Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis typically affecting multiple organ systems. We report 2 patients who presented with homonymous hemianopia and were ultimately diagnosed with biopsy-confirmed ECD. We review the spectrum of ECD and its treatment as well as histopathological and immunohistochemical differentiation from other histiocytic disorders.	24743792
2014 Apr 13	Neurologia	Neurological manifestations in Erdheim-Chester disease: Two case reports	Rouco I, Arostegui J, Cánovas A, González Del Tánago J, Fernández I, Zarranz JJ	Servicio de Neurología, Hospital Universitario de Cruces, Osakidetza, Baracaldo, Vizcaya, España idoia.rouco@gmail.com	[In Spanish]	24735941
2014 Apr 3	Pediatr Neurosurg.	Cerebral Erdheim-Chester Disease Mimicking High-Grade Glial Tumor: A Case Report.	Ekşi MS, Demirci Otluoğlu G, Uyar Bozkurt S, Sav A, Bayri Y, Dağçınar A.	Department of Neurosurgery, Marmara University Medical School, Istanbul, Turkey	Erdheim-Chester disease (ECD) is a non-Langerhans histiocytosis. It may present in every organ in the body, but isolated central nervous system involvement, especially a supratentorial intra-axial location, is extremely rare. We present a case of ECD of supratentorial intra-axial origin and discuss the clinical presentation, diagnosis and management strategies.	24713668

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Apr 2	Ann Vasc Surg	Bilateral Renal Artery Involvement of Erdheim-Chester Disease.	Yaeger AA, Weaver FA, Woo K.	Division of Vascular Surgery and Endovascular Therapy, Keck School of Medicine, University of Southern California, Los Angeles, CA. Electronic address: karen.woo@surgery.usc.edu	To report a rare case of nonatherosclerotic renal artery disease (NARAD) in a patient with Erdheim-Chester disease (ECD), a non-Langerhans cell histiocytosis. The patient is a 75-year-old male who presented with 2 years of hypertension secondary to right renal artery occlusion incidentally found on cardiac catheterization. Open bilateral renal revascularization was performed because of improved long-term graft patency demonstrated in other forms of NARAD. Nine months postoperatively, the patient's hypertension was significantly improved and surveillance duplex demonstrated patent bilateral renal artery bypasses. Revascularization in patients with NARAD, such as ECD, should be managed in an open approach because of the superior long-term graft patency and blood pressure control in a disease where progression to vessel fibrosis and atherosclerosis could interfere with endovascular stent patency.	24704054
2014 Mar 26	Ann Rheum Dis	An ophthalmologic diagnostic error leading to a rare systemic diagnosis: Erdheim-Chester disease	Gilles M, Alberti N, Seguy C, Leger F, Frulio N, Pechmeja J, Longueville E, Korobelnik JF.	Service d'ophtalmologie, centre hospitalier Pellegrin, place Amélie-Raba-Léon, 33000 Bordeaux, France. Electronic address: marionkgilles@gmail.com.	INTRODUCTION: Erdheim-Chester disease is a rare systemic disease. The diagnosis is difficult due to significant clinical and morphological polymorphism. Orbital involvement is rare, but constitutes a classic means of detection. OBSERVATION: We report the case of a 60-year-old man, who consulted for evaluation of bilateral retro-orbital tumors. These tumors had been discovered on head CT two years previously during work-up of proptosis. Two biopsies were performed. The first one revealed polymorphous inflammatory tissue. The second one revealed intense granulomatous reaction, rich in non-specific foamy histiocytes. Thoracic-abdominal-pelvic CT scan detected peri-aortic and retroperitoneal infiltration. The association of these signs pointed to a diagnosis of Erdheim-Chester disease, confirmed by the re-examination of the histological samples. DISCUSSION: Erdheim-Chester disease is a rare non-Langerhans histiocytosis with a specific tropism for perivascular and fatty connective tissue. The cause is not known. The diagnosis of this systemic disease is histological. CONCLUSION: In the case of bilateral intra-orbital tumors, the diagnosis of Erdheim-Chester disease must be considered.	24674299

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Mar 26	Ann Rheum Dis.	BRAFV600E-mutation is invariably present and associated to oncogene-induced senescence in Erdheim-Chester disease.	Cangi MG, Biavasco R, Cavalli G, Grassini G, Dal-Cin E, Campochiaro C, Guglielmi B, Berti A, Lampasona V, von Deimling A, Sabbadini MG, Ferrarini M, Doglioni C, Dagna L.	Unit of Pathology, IRCCS San Raffaele Scientific Institute, Milan, Italy; lorenzo.dagna@unisr.it	OBJECTIVES: Erdheim-Chester disease (ECD) is a rare form of histiocytosis characterised by uncontrolled chronic inflammation. The oncogenic BRAFV600E mutation has been reported in biopsies in 19 out of 37 patients with ECD from the largest published cohort, but never found in the patients' peripheral blood. Also, the role of the mutation in the pathogenesis of the disease has not been elucidated yet. BRAFV600E has been associated with oncogene-induced senescence (OIS), a protective mechanism against oncogenic events, characterised by the induction of proinflammatory pathways. METHODS: We verified the BRAF status in biopsies and peripheral blood from 18 patients with ECD from our cohort and matched controls by means of immunohistochemistry and of an ultrasensitive assay, based on the combination of a locked nucleic acid PCR and pyrosequencing. Droplet digital PCR was used to confirm the findings. We also evaluated the presence of senescence markers in ECD histiocytes. RESULTS: BRAFV600E mutation was present in all the biopsy and peripheral blood samples from patients with ECD and in none of the controls. ECD histiocytes and a fraction of circulating monocytes from patients with ECD showed signs of a constitutive activation of the MAPK pathway. Moreover, BRAF-mutated histiocytes expressed markers of OIS. CONCLUSIONS: The oncogenic BRAFV600E mutation is present in biopsies and in the peripheral blood from all patients with ECD who were evaluated and is associated with OIS. These findings have significant implications for the pathogenesis, diagnosis and treatment of ECD.	24671772
2014 Feb 6	J Clin Neurosci.	Mistaken identity: Granular cell astrocytoma masquerading as histiocytosis of the central nervous system.	Campbell RN, Liew MS, Gan HK, Cher L.	Joint Austin-Ludwig Oncology Unit, Level 4 Olivia Newton-John Cancer & Wellness Centre, Austin Hospital, 145 Studley Road, Heidelberg, VIC 3084, Australia; Neurology Unit, Austin Health, VIC, Australia. Electronic address: lmcher@mac.com	Granular cell astrocytoma (GCA) is an uncommon malignant glial tumour that is associated with a poor prognosis. GCA cells have some morphological and immunohistochemical similarities to macrophages. In this case, a small biopsy contained no typical astrocytoma and large rounded lesional cells were interpreted as negative for glial fibrillary acidic protein and S100 and positive for CD68, a commonly used marker for macrophages. A diagnosis of a histiocytosis was made. When the patient failed to respond to first and second line therapy, tumour resection was undertaken and the pathology then showed typical morphologic and immunohistochemical features of glioblastoma (astrocytoma World Health Organization grade IV).	24594450

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Apr	Curr Rheumatol Rep	Erdheim-chester disease	Haroche J, Arnaud L, Cohen-Aubart F, Hervier B, Charlotte F, Emile JF, Amoura Z.	Department of Internal Medicine and French Reference Center for Rare Autoimmune and Systemic Diseases, Assistance Publique-Hôpitaux de Paris, Pitié-Salpêtrière Hospital, Paris, France, Email: julien.haroche@psl.aphp.fr	Erdheim-Chester disease (ECD) is a rare (approximately 500 known cases worldwide), non-inherited, non-Langerhans form of histiocytosis of unknown origin, first described in 1930. It is characterized by xanthomatous or xanthogranulomatous infiltration of tissues by foamy histiocytes, "lipid-laden" macrophages, or histiocytes, surrounded by fibrosis. Diagnosis of ECD involves the analysis of histiocytes in tissue biopsies: these are typically foamy and CD68+ CD1a- in ECD, whereas in Langerhans cell histiocytosis (LCH) they are CD68+ CD1a+. (99)Technetium bone scintigraphy revealing nearly constant tracer uptake by the long bones is highly suggestive of ECD, and a "hairy kidney" appearance on abdominal CT scan is observed in approximately half of ECD cases. Central nervous system involvement is a strong prognostic factor and an independent predictor of death in cases of ECD. Optimum initial therapy for ECD seems to be administration of interferon α (or pegylated interferon α), and prolonged treatment significantly improves survival; however, tolerance may be poor. Cases of ECD present with strong systemic immune activation, involving IFNα, IL-1/IL1-RA, IL-6, IL-12, and MCP-1, consistent with the systemic immune Th-1-oriented disturbance associated with the disease. More than half of ECD patients carry the BRAF (V600E) mutation, an activating mutation of the proto-oncogene BRAF. A small number of patients harboring this mutation and with severe multisystemic and refractory ECD have been treated with vemurafenib, a BRAF inhibitor, which was proved very beneficial.	24532298
2014 Feb 16	Endocrine	Surgical biopsies in patients with central diabetes insipidus and thickened pituitary stalks	Jian F, Bian L, Sun S, Yang J, Chen X, Chen Y, Ma Q, Miao F, Wang W, Ning G, Sun Q.	Department of Endocrine and Metabolic Diseases, Ruijin Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai, 200025, China. Email: rjns123@163.com	Thickened pituitary stalks (TPSs) on magnetic resonance imaging (MRI) result from diverse pathologies; therefore, it is essential to make specific diagnoses for clinical decision-making. The diagnoses and indications for surgical biopsies in patients with central diabetes insipidus (CDI) and TPSs are thoroughly discussed in this paper. Thirty-seven patients with CDI and TPSs were retrospectively reviewed. The mean age at the diagnosis of CDI was 29.0 ± 15.9 years (range 8.0-63.3), and the median duration of follow-up was 5.5 ± 2.8 years (range 0.7-13.0). Anterior pituitary hormone deficiencies were documented in 26 (70.3 %) patients. All patients had a TPS on MRI at the diagnosis of CDI, and 21 (56.8 %) patients exhibited radiological changes during the follow-up. Of these 21 patients, 11 exhibited increases in the thickness of the stalk, and two patients exhibited reversals of the TPSs. Involvements of the hypothalamus, pituitary gland, basal ganglia or supersellar, and pineal gland were found in four, three, one, and 1 patient, respectively. Ultimately, clear diagnoses were established in 17 patients who underwent biopsies, nine of whom had germinomas, six of whom had Langerhans cell histiocytosis, one of whom had a granular cell tumor, and one of whom had Erdheim-Chester disease. Patients with CDI and TPSs should submit to periodic clinic follow-ups with serial MRI assessments to establish anterior pituitary deficiencies and to detect radiological progressions that are appropriate for surgical biopsies. Endoscopic-assisted microsurgery via the supraorbital keyhole approach is a good choice for the biopsy of pituitary stalk lesions.	24532100

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2014 Feb 16	Jpn J Radiol	Erdheim-Chester disease with an 18F-fluorodeoxyglucose-avid breast mass and BRAF V600E mutation	Furuta T, Kiryu S, Yamada H, Hosoi M, Kurokawa M, Morikawa T, Shibahara J, Ohtomo K.	Department of Radiology, Research Hospital, The Institute of Medical Science, The University of Tokyo, 4-6-1, Shirokanedai, Minato-ku, Tokyo, 108-8639, Japan, Email: tfuruta-tky@umin.ac.jp	Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytosis. Herein we report a case of a 49-year-old woman who developed bilateral knee pain. Imaging procedures revealed multiple long bone lesions and a well-defined 18F-fluorodeoxyglucose-avid mass in the left breast. The breast mass was resected, and an open biopsy was performed on the right femoral lesion. Both specimens revealed involvement by histiocytic infiltrates with features suggestive of ECD. The BRAF V600E mutation was detected by DNA sequencing and immunohistochemistry.	24531980
2014 Feb 10	Acta Otorrinolaringol Esp	Erdheim-Chester disease in a sinonasal location	Nazar R, Ortega G, Miranda G, Naser A.	Departamento de Otorrinolaringología, Hospital Clínico de la Universidad de Chile, Santiago de Chile, Chile. Email: rnazars@gmail.com	Not available.	24524800
2014 Feb 1	Blood Cells Mol Dis	BRAF - A new player in hematological neoplasms	Machnicki MM, Stoklosa T.	Department of Immunology, Medical University of Warsaw, Banacha 1A, 02-097 Warsaw, Poland. Email: tomasz.stoklosa@wum.edu.pl	BRAF oncogenic kinase has become a target for specific therapy in oncology. Genetic characterization of a predominant V600E mutation in melanoma, thyroid cancer, and other tumors became a focus for developing specific inhibitors, such as vemurafenib or dabrafenib. Our knowledge regarding the role of mutated BRAF in hematological malignancies has grown quickly as a result of new genetic techniques such as next-generation sequencing. This review summarizes current knowledge regarding the role of BRAF in lymphoid and myeloid neoplasms, with a focus on hairy-cell leukemia, Langerhans cell histiocytosis, and Erdheim-Chester disease.	24495477
2014 Jan 27	Arthritis Care Res (Hoboken)	New insights in pathogenesis of Erdheim-Chester disease	Bachmeyer C, Buffo M, Soyez B.	Service de Médecine Interne, Hôpital Tenon (AP-HP), 75020, Paris, France Email: claude.bachmeyer@tnn.aphp.fr	Not available.	24469999
2014 Jan 27	Arthritis Care Res (Hoboken)	Vemurafenib in Erdheim-Chester disease	Santos-Ortega A, Arcos-Bertiz IL, Martinez-Valle F.	Research Unit in Systemic Autoimmune Diseases, Internal Medicine Department, Cardiology Department, Vall D'hebron Hospital, Barcelona, Spain Email: ferranmartinezvalle@gmail.com	Not available.	24469977

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Dec 21	Clin Imaging	Pineal gland involvement in Erdheim-Chester disease detected on 18F-FDG PET-CT imaging: a case report and review of literature	Mukherjee A, Dhull VS, Karunanithi S, Sharma P, Durgapal P, Kumar R.	Department of Nuclear Medicine, All India Institute of Medical Sciences, New Delhi, India Email: rkphulia@yahoo.com	Erdheim-Chester disease (ECD) is a rare non-Langerhan's cell histiocytosis affecting multiple organ systems. The most common systemic manifestations are bone lesions, infiltration of the pituitary stalk sometimes leading to diabetes insipidus, pulmonary fibrosis, cardiac failure and exophthalmus. Neurological symptoms as the first clinical manifestations of ECD have been reported in less than one third of cases. We report a rare presentation of a patient of ECD on 18F-fluorodeoxyglucose (18F-FDG) positron emission tomography-computed tomography which revealed abnormal 18F-FDG accumulation in the region of pineal gland, pericardium and bilateral distal tibiae.	24461468
2014 Jan 15	Dermatol Online J	Anakinra-responsive lichen planus in a woman with Erdheim-Chester disease: a therapeutic enigma	Cohen PR, Kurzrock R.	University of California San Diego 3855 Health Sciences Drive, #3069 La Jolla, CA 92093 USA	Background: Anakinra is a recombinant form of interleukin-1 receptor antagonist. It is the drug of choice for Schnitzler syndrome and cryopyrin-associated periodic syndromes. It has also recently been demonstrated to have activity in the treatment of the non-Langerhans cell histiocytosis known as Erdheim-Chester disease. Purpose: To describe the activity of anakinra in a patient with co-existing lichen planus and Erdheim-Chester disease. Methods: A 43-year-old woman with progressive Erdheim-Chester disease presented for management of her night sweats and chills, systemic skeletal bone pain, and neurologic (diabetes insipidus) manifestations. She also had widespread cutaneous lichen planus. Anakinra, 100 mg subcutaneously daily, was initiated for the treatment of her Erdheim-Chester disease. Results: Within 2 days of starting anakinra, there was prompt resolution of her Erdheim-Chester disease-related symptoms. Subsequently, her bone pain resolved and her diabetes insipidus improved. Also, the lichen planus-associated pruritus rapidly ceased and most of the skin lesions improved. Conclusions: Our experience confirms the efficacy of anakinra for the treatment of Erdheim-Chester disease. The concomitant improvement of her lichen planus on anakinra suggests that this agent warrants additional study in this disorder.	24456945
2014 Jan	Intern Med J	Successful treatment of Erdheim-Chester disease with combination of interleukin-1-targeting drugs and high-dose glucocorticoids	Darstein F, Kirschey S, Heckl S, Rahman F, Schwarting A, Schuchmann M, Galle PR, Zimmermann T.	Tim Zimmermann, Universitätsmedizin Mainz, Langenbeckstraße 1, Mainz 55131, Germany. Email: tim.zimmermann@unimedizin-mainz.de	Erdheim-Chester disease (ECD) is a rare histocytic disorder. We report a case of a 45-year-old male ECD patient with severe clinical manifestation (urinary obstruction due to retroperitoneal mass with hydronephrosis, involvement of long bones) and central nervous system involvement (hemiparesis, aphasia and diabetes insipidus). Diagnosis was confirmed by typical clinical, radiological and histological findings. Under immunosuppressive therapy with prednisolone and interleukin-1A receptor antagonist (Anakinra, Kineret, Swedish Orphan Biovitrum AB, Stockholm, Sweden), a rapid improvement of the patients' symptoms and condition was observed. This is the first report of a successful combination therapy of Anakinra and glucocorticoids. Furthermore, current literature about ECD and treatment options are discussed.	24450524
2014 Jan 14	Clin Exp Rheumatol	Imaging in Erdheim-Chester disease: classic features and new insights	Moulis G, Sailler L, Bonneville F, Wagner T.	Internal Medicine Department, Toulouse University Hospital, UMR INSERM-UPS 1027, University of Toulouse, Toulouse, France. guillaume.moulis@univ-tlse3.fr	Erdheim-Chester disease is a rare non-Langerhans cell histiocytosis. Osseous involvement is the most frequent feature with bilateral and symmetric osteosclerotic changes in long bone diaphyseal and metaphyseal regions, classically sparing epiphyses. 99mTc scintigraphy shows bilateral and symmetrical metaphysal and diaphyseal increased uptake in almost all the patients, even asymptomatic. Other classical features on CT-scan, very evocative of Erdheim-Chester disease, must be recognised: e.g. 'coated' aorta, 'hairy kidney' patterns. New imaging techniques such as MRI have led to a better description of cardiac and central nervous system involvements. Pachymeningitis and right atrium wall infiltration are new evocative images of this disease. Fluorodeoxyglucose Positron Emission Tomography in the diagnosis or prognosis assessment is still discussed. The objective of this review is to discuss the place of each imaging technique in Erdheim-Chester disease in 2013.	24428974

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Nov-Dec	G Ital Nefrol	Renal involvement in erdheim-chester disease	Li Cavoli G, Bono L, Tortorici C, Ferrantelli A, Zagarrigo C, Servillo F, Giammarresi C, Tralongo A, Schillaci O, Coglitore M, Rotolo U.	Department of Nephrology and Dialysis, Civic and Di Cristina Hospital, Palermo, Italy	The Erdheim-Chester disease is a rare form of Langherans cells. Since 1987 it is distinguished from other istiocytosis previously identified. The diagnosis of the disease relies on defined radiological (bone imaging) and pathological (histiocytic infiltration) criteria. Bone disease is crucial but systemic manifestations are reported more frequently at onset. Renal involvement is always asymptomatic at onset of disease or in the follow-up. In this review we analyze the reports of the literature; we highlight 3 pathological mechanisms of renal involvement: renal and retroperitoneal infiltration, urinary tract obstruction, renal arteries stenosis. No treatment to date has demonstrated an improvement in survival of patients with EC. Renal involvement is therefore symptomatic (ureteral stenting, percutaneous nephrostomy) or is adopted a wait-and-see attitude. [Article in Italian.]	24402664
2013 Dec 24	Skeletal Radiol	Erdheim-Chester disease: An unusual presentation of an uncommon disease	Bindra J, Lam A, Lamba R, Vanness M, Boutin RD.	Department of Radiology, University of California, Davis, School of Medicine, Sacramento, CA, USA, jasjeet.bindra@ucdmc.ucdavis.edu	Erdheim-Chester disease (ECD) is a rare, non-Langerhans cell histiocytosis with classic radiographic findings of patchy or diffuse osteosclerosis predominantly involving the long bones in a bilaterally symmetrical pattern. A 49-year-old woman presented with diffuse lymphadenopathy, painful skin lesions, and constitutional symptoms. Recent history was significant for a nontraumatic fracture of the tibia 3 weeks prior to admission. Physical examination and laboratory studies were notable for lower extremity pain and swelling, nodular lesions on the skin, and normocytic, normochromic anemia. Plain radiographs showed a lytic pattern of destruction with a superimposed fracture in the left proximal tibia. MRI showed focal bone marrow replacement extending from the subchondral bone to the tibial diaphysis. Excisional lymph node and skin biopsies of the lesions demonstrated a CD-68 positive, S-100 variable, and CD1a-negative histiocytic cell proliferation filling the dermis and completely replacing the sampled lymph node with an accompanying chronic inflammatory infiltrate and fibrosis, pathognomonic for ECD. We report an unusual case of ECD presenting initially as diffuse, painful lymphadenopathy, and subsequently demonstrating a lytic lesion of the tibia underlying a nontraumatic fracture.	24366632
2013 Sep-Oct	Tumori	Salvage lenalidomide in four rare oncological diseases	Szturz P, Adam Z, Rehak Z, Koukalova R, Kren L, Moulis M, Krejcí M, Mayer J.	Petr Szturz, MD, PhD, Department of Internal Medicine, Hematology and Oncology, University Hospital Brno, Jihlavská 20, 625 00 Brno, Czech Republic. Tel +420-5-32232934; fax +420-5-32233603; email petr.szturz@fnbrno.cz	In rare disorders, there are often no standard therapy recommendations. Patients with refractory disease may require novel experimental approaches. Applied as second- up to fourth-line treatment, lenalidomide (10-25 mg perorally on days 1-21 in a 28-day cycle) was used in our cohort of four adult patients with aggressive, multisystem and relapsing diseases. Complete and long-lasting remissions (more than 1 year, no maintenance therapy) were achieved in patients with Langerhans cell histiocytosis (11 cycles, combination with dexamethasone and etoposide, consolidated by allogeneic blood stem cell transplant) and plasma-cell Castleman disease (15 cycles, monotherapy). Mixed response with complete disappearance of brain infiltrates was reached in Erdheim-Chester disease (6 cycles, monotherapy) and gastrointestinal bleeding was well controlled in multiple angiomatosis (9 cycles, combination with thalidomide). For disease activity evaluation each patient underwent fluorine-18-fluorodeoxyglucose positron emission tomography/computed tomography scan imaging, which was complemented by clinical and laboratory investigations.	24362880

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Dec	Recenti Prog Med	Cardiovascular involvement in Erdheim-Chester syndrome: clinical and therapeutic implications	Lauretta L, Dagna L, Alberti L, Loiacono F, De Cobelli F, Sanvito F, Moriggia S, Margonato A, Fragasso G.	Cardio-Thoracic-Vascular Department, San Raffaele Scientific Institute, Milan, Italy email: lorenzo.dagna@unisr.it	Erdheim-Chester disease (ECD) is a rare form of non-Langerhans' cell histiocytosis of unknown etiology and its incidence is constantly increasing. ECD is characterized by a xantomatous or xanthogranulomatous infiltration of various tissues by foamy histiocytes surrounded by fibrosis. ECD is characterized by multi-organ involvement and is generally associated with a poor prognosis with a median survival of 32 months after diagnosis. Cardiovascular involvement concerns mainly the thoraco-abdominal aorta and pericardium. Less frequently, infiltration affects the myocardial tissue, especially the right atrium, and the valvular endocardium. Recently, the involvement of the vena cava has also been described. The diagnosis of ECD is made by the identification of foamy histiocytes CD68 positive and CD1a/S100 negative embedded in a polymorphic inflammatory tissue on biopsy. Despite the adoption of several therapeutic strategies until now prognosis has remained poor. Interferon-α can be considered the first line therapy, but its effects on central nervous system and cardiovascular localization have been shown to be often poor. In this context a combined treatment with the anti-TNFα monoclonal antibody infliximab and methotrexate seems to be effective and well tolerated.	24362833
2013 Oct-Dec	J Postgrad Med	Erdheim-Chester disease: A clinical and radiological masquerade	Wadhwa V, Marcus RW, Carrino JA, Chhabra A.	The Russell H. Morgan Department of Radiology and Radiological Science, Johns Hopkins University School of Medicine, Baltimore	No abstract available.	24346395
2013 Dec 3	Endocr J	Erdheim-Chester disease and pituitary involvement: a unique case and the literature	Manaka K, Makita N, Iiri T.	Department of Endocrinology and Nephrology, The University of Tokyo School of Medicine, Tokyo 113-8655 Japan E-mail: norimaki-tky@umin.ac.jp	An early thirties man diagnosed with Erdheim-Chester disease (ECD) was simultaneously disclosed to have hypogonadotropic hypogonadism, central adrenal insufficiency, and GH deficiency in addition to central diabetes insipidus (CDI). Pituitary magnetic resonance imaging (MRI) showed swelling in the stalk, enlargement of the anterior lobe with delayed enhancement, and loss of high intensity of the posterior lobe on T1-weighted images, suggesting of pituitary involvement of ECD. Three months after starting treatment with interferon α and zoledronic acid, polyuria and polydipsia were ameliorated without DDAVP, accompanied with improvement of MRI. Simultaneously technetium-99m bone scintigraphy showed improvement, accompanied with a relief of bone pain and high fever. In contrast, he developed secondary hypothyroidism with slight enlargement of anterior pituitary gland without relapse of CDI, suggesting of different responses to treatment with interferon α between anterior pituitary lobe and posterior one. So far he continues to be replaced with deficient hormone replacement therapy. As for bone pain, it remains to be controlled with the decreased levels of bone resorption marker with decreased abnormal uptake in bone scintigraphy although zoledronic acid was discontinued for osteonecrosis of the jaw. For four years, he has not showed new involvement at other organs besides bones and the pituitary. While CDI is known to be very common in ECD, improvement of CDI has been reported in a few cases. Other endocrine manifestations, especially with detailed endocrine status, have been also reported in limited cases. Thus we report this case and review the literature.	24304810

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Nov	Informa Healthcare	Strategies and treatment alternatives in the management of Erdheim–Chester disease	Roei David Mazor, Mirra Manevich-Mazor, & Yehuda Shoenfeld	Yehuda Shoenfeld Sheba Medical Center, Tel Hashomer, Israel; shoenfel@post.tau.ac.il	**Introduction:** The treatment of Erdheim–Chester disease (ECD) remains a challenging task. Current opinion corroborates interferon-α as the first-line treatment. Several second-line treatments exist for patients who advance on or who do not tolerate interferon-α. Among them are vemurafenib, anakinra, infliximab and cladribine. **Areas covered**: A systematic search of PubMed was employed to identify all the articles relating to the treatment of ECD. The quintessential facts are as follows: Interferon-α increases survival among ECD patients at dosages of as low as 3 × 106 IU × 3/week. Nevertheless, the more resilient cardiovascular and central nervous system (CNS) disease foci necessitate dosage regimens of as high as 9 × 106 IU × 3/week. Anakinra, administered at dosages of 1 – 2 mg/kg/day, should be reserved for patients with mild disease. Infliximab, administered at a dosage of 5 mg/kg/6 weeks induced regression of ECD-related cardiovascular lesions. Vemurafenib, administered at a dosage of 960 mg/day, induced remarkable improvement in the symptoms, CRP levels and PET findings of patients harboring the V600E BRAF mutation. Cladribine may be effective administered at dosages of 0.07 – 0.14 mg/kg/day for five consecutive days. However, it should be reserved for patients with moderate–to-severe disease who failed on or who are not candidates for other second-line treatments. **Expert opinion:** Recent advancements in the recognition of biological targets imbued the therapeutic repertoire of ECD with novel personally tailored treatments. As the zealous hunt for future remedies persists, one must always remember that the success of an ongoing treatment is congruent with an assured, well-informed patient.	
2013 Nov 15	Scand J Infect Dis	Erdheim-Chester disease: A rare cause of recurrent fever of unknown origin mimicking lymphoma	Mariampillai A, Sivapiragasam A, Kumar A, Hindenburg A, Cunha BA, Zhou J	B. A. Cunha, Infectious Disease Division, Winthrop-University Hospital, Mineola, NY 11501; bacunha@winthrop.org	We report the case of a patient with recurrent fever of unknown origin (FUO) with prominent back pain, hepatosplenomegaly, and abdominal/pelvic adenopathy suggesting lymphoma. A bone biopsy showed histiocytic infiltration. Studies for lymphoma were negative, but immunohistochemical stains were diagnostic of Erdheim-Chester disease (ECD). ECD should be included as a rare cause of recurrent FUO with bone involvement.	24228820
2013 Oct 7	J Clin Ultrasound.	Virtual CT sonographically guided biopsy of a retroperitoneal mass in a patient with Erdheim-Chester disease	Tombesi P, Di Vece F, Rinaldi R, Ermili F, Sartori S.	Section of Interventional Ultrasound, Department of Internal Medicine, St. Anna Hospital, Ferrara, Italy.	Image fusion between sonography and CT allows real-time synchronization of CT multiplanar reconstructed images with the corresponding sonographic images. This technique has mainly been used in liver imaging. We report the use of image fusion to target and successfully guide the percutaneous biopsy of a retroperitoneal a mass. This technique represents a promising tool in abdominal imaging, and it should be considered for the biopsy of lesions that are difficult to approach with conventional imaging guidance techniques. © 2013 Wiley Periodicals, Inc. J Clin Ultrasound, 2013;	24123060
2013 Oct	Hong Kong Med J	Erdheim-Chester disease: an uncommon cause of upper urinary tract obstruction	Tsu JH, Yuen SK, Cheung H, Lee YW, Liu PL.	Department of Surgery, Caritas Medical Centre, 111 Wing Hong Street, Shamshuipo, Kowloon, Hong Kong; thl589@ha.org.hk	Erdheim-Chester disease is a rare non-Langerhans form of systemic histiocytosis of unknown origin. We describe a 45-year-old man presenting with bilateral hydronephrosis suggestive of extrinsic urinary tract obstruction. Computed tomography revealed extensive hypodense soft tissue infiltration in the retroperitoneum surrounding the kidneys. Needle biopsy of the retroperitoneal soft tissue revealed aggregates of lipid-laden histiocytes expressing CD68 but negative for CD1a and S100 protein. The diagnosis of Erdheim-Chester disease was supported by typical radionuclide bone scinitigraphic findings. Treatment with prednisolone, sirolimus, and regular ureteric stent revision was initiated to achieve adequate urinary tract drainage. To our knowledge, this is the second patient with Erdheim-Chester disease reported in Hong Kong. A high index of suspicion is required to avoid delay in the diagnosis of this rare disease.	24088590

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013	Adv Immunol	Pathological consequence of misguided dendritic cell differentiation in histiocytic diseases	Berres ML, Allen CE, Merad M.	Department of Oncological Sciences, Icahn School of Medicine at Mount Sinai, New York, USA; Tisch Cancer Institute; Immunology Institute; marie.berres@mssm.edu	Histiocytic disorders represent a group of complex pathologies characterized by the accumulation of histiocytes, an old term for tissue-resident macrophages and dendritic cells. Langerhans cell histiocytosis is the most frequent of histiocytosis in humans and has been thought to arise from the abnormal accumulation of epidermal dendritic cells called Langerhans cells. In this chapter, we discuss the origin and differentiation of Langerhans cells and dendritic cells and present accumulated evidence that suggests that Langerhans cell histiocytosis does not result from abnormal Langerhans cell homeostasis but rather is a consequence of misguided differentiation programs of myeloid dendritic cell precursors. We propose reclassification of Langerhans cell histiocytosis, juvenile xanthogranuloma, and Erdheim-Chester disease as inflammatory myeloid neoplasias.	24070383
2013 Oct	Int J Hematol	Erdheim-Chester disease and Schnitzler syndrome: so near, and yet so far	Szturz Petr, Hlavatý L, Prášek J, Dvořáková D.	Department of Internal Medicine, Hematology and Oncology, School of Medicine, University Hospital Brno and Masaryk University, Jihlavská 20, 625 00, Brno, Czech Republic, petr.szturz@fnbrno.cz	No abstract available.	24046179
2013	Bulletin of the Hospital for Joint Diseases	Erdheim-Chester disease--clinical pathological case discussion	Rivera TL, Irish RD, Hoda SA, Steiner GC, Rackoff PJ, Fischer HD	Tania L. Rivera, M.D., Department of Medicine, Division of Rheumatology, Beth Israel Medical Center, University Hospital for the Albert Einstein College of Medicine, 350 East 17th Street, Baird Hall Suite 20BH54, New York, New York 10003; tarivera@chpnet.org	(See http://www.ncbi.nlm.nih.gov/corehtml/query/egifs/http:--www.nyuhjdbulletin.org-icon.jpg for a the full article.)	24032617

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Sept 8	Orphanet Journal of Rare Diseases	Erdheim-Chester Disease: a comprehensive review of the literature	Roei D Mazor, Mirra Manevich-Mazor, Yehuda Shoenfeld	The Laura Schwarz-Kipp Chair for Research of Autoimmune Diseases, Tel-Aviv University, Tel-Aviv, Israel rd.mazor@gmail.com, mirra.manevich@gmail.com, shoenfel@post.tau.ac.il	Erdheim-Chester Disease (ECD) is a rare form of non Langerhans' cell histiocytosis. Individuals affected by this disease are typically adults between their 5th and 7th decades of life. Males and females are almost equally affected. The multi systemic form of ECD is associated with significant morbidity, which may arise due to histiocytic infiltration of critical organ systems. Among the more common sites of involvement are the skeleton, central nervous system, cardiovascular system, lungs, kidneys (retroperitoneum) and skin. The most common presenting symptom of ECD is bone pain. The etiology of ECD is unknown yet thought to be associated with an intense TH1 immune response. It may also be associated with the V600E BRAF mutation, as described in as many as half of the patients in recent studies. Bilateral symmetric increased tracer uptake on 99mTc bone scintigraphy affecting the periarticular regions of the long bones is highly suggestive of ECD. However, definite diagnosis of ECD is established only once CD68(+), CD1a(−) histiocytes are identified within a biopsy specimen. At present, this obscure ailment embodies numerous challenges to medical science. Given its rarity, it is diagnostically elusive and requires a high level of clinical suspicion. Therapeutically, it is of limited alternatives. Currently, interferon-α is the most extensively studied agent in the treatment of ECD and serves as the first line of treatment. Treatment with other agents is based on anecdotal case reports and on the basis of biological rationale. Nevertheless, cladribine (2CDA), anakinra and vemurafenib are currently advocated as promising second line treatments for patients whose response to interferon α is unsatisfactory. Overall, the 5 year survival of ECD is 68%. Herein, the authors mustered and brought about a panoramic consolidation of all the relevant facts regarding ECD. This work highlights the different clinical, radiological and pathological manifestations associated with ECD, the differential diagnoses, the various treatment options and the acknowledged science explaining the disease.	24011030
2013 Aug 13	Joint Bone Spine	Successful treatment of Erdheim-Chester disease by interleukin-1 receptor antagonist protein	Courcoul A, Vignot E, Chapurlat R.	Inserm UMR1033, université de Lyon, Hospices Civils de Lyon, Department of rheumatology, Pavillon F, hospital Edouard-Herriot, 5, place d'Arsonval, 69003 Lyon, France roland.chapurlat@chu-lyon.fr	Erdheim-Chester disease is a rare non-langerhans systemic histiocytosis of unknown origin, associated with bone diseases and severe visceral complications. Therapies have been disappointing. A recombinant form of interleukin-1 receptor antagonist (anakinra) has been used in a few cases when usual treatment fails. We report a new case of successfully interleukin-1 receptor antagonist treatment in Erdheim-Chester disease.	23953221
2013 Aug 8	Blood	Detection of an NRAS mutation in Erdheim-Chester disease	Diamond EL, Abdel-Wahab O, Pentsova E, Borsu L, Chiu A, Teruya-Feldstein J, Hyman DM, Rosenblum M.	Department of Neurology, Memorial Sloan-Kettering Cancer Center, New York, NY diamone1@mskcc.org	No abstract available.	23929840

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Jul 26	Rheumatology (Oxford)	TNF-α in Erdheim-Chester disease pericardial effusion promotes endothelial leakage in vitro and is neutralized by infliximab	Ferrero E, Belloni D, Corti A, Doglioni C, Dagna L, Ferrarini M. ferrero.elisabetta@hsr.it	San Raffaele Scientific Institute, Milan, Italy and Vita-Salute San Raffaele University, Milan, Italy	No abstract available.	23893523
2013 Jul	Neurology	Teaching NeuroImages: Ataxia and diabetes insipidus	Lefaucheur R, Maltête D, Haroche J, Borden A, Wallon D, Bourre B.	Department of Neurology, Rouen University Hospital and University of Rouen, Rouen; Assistance Publique-Hôpitaux de Paris, Pitié-Salpêtriére Hospital, Paris, France romain.lefaucheur@chu-rouen.fr	A 63-year-old woman presented with recent cerebellar ataxia. Diabetes insipidus had been diagnosed 35 years earlier. Brain MRI showed fluid-attenuated inversion recovery hyperintensities involving cerebellar peduncles and pons, with punctiform gadolinium enhancement on T1 sequences (figure, A). Leg X-rays revealed cortical osteosclerosis of both tibias (figure, B). Tibia biopsy revealed bone remodeling associated with bone marrow fibrosis and lymphohistiocytic reaction confirming the diagnosis of Erdheim-Chester disease (ECD).	23858417
2013 Jun	J Am Acad Dermatol	Diabetes insipidus, bone lesions, and new-onset red-brown papules in a 42-year-old man	Kornik RI, Naik HB, Lee CC, Estrada-Veras J, Gahl WA, Cowen EW.	Dermatology Branch, Center for Cancer Research, National Cancer Institute, National Institutes of Health, Bethesda, Maryland 20892, USA; Juvianee.EstradaVeras@nih.gov	No abstract available.	23466246
2013 Jun 21	Dig Liver Dis.	Portal hypertension and ascites secondary to Erdheim Chester Disease without intrinsic liver involvement on liver biopsy	Tsynman DN, Weaver C, Taboada S, Findeis-Hosey J, Maliakkal B, Huang J.	University of Rochester, Department of Gastroenterology and Hepatology, United States. Electronic address: Donald_Tsynman@URMC.Rochester.edu	No abstract available.	23796550
2012 Nov	West Indian Med J.	Erdheim-Chester disease in a child	Wen C, Liang Q, Yi Z, Wan W.	Department of Paediatrics, Xiangya Second Hospital of Central South University, Changsha City, China. zz040217@yahoo.com.cn	Erdheim-Chester disease (ECD) is a rare systemic non-Langerhans histiocytosis that affects multiple organ systems. It occurs more often in adults, and paediatric ECD is extremely rare. The diagnosis of ECD can be established based on clinical presentations and imaging but the final diagnosis should be based on biopsy. Treatment of ECD has involved the use of corticosteroids, radiotherapy, chemotherapy, surgery and haematopoietic stem cell transplantation, yet the efficacy of these treatments is difficult to determine. At present, it is thought that the treatment of interferon-alpha (IFN-alpha) is safe and effective for ECD. Herein, we report on an 11-year old girl who was admitted to hospital because of systemic bone pain and limping, and the final diagnosis of ECD was based on evidence provided by her clinical presentation, imaging and biopsy of a lesion of the right ilium. The patient was treated with subcutaneous IFN-alpha at a dosage of 3 x 10(6) units three times weekly for 19 months. We thought that the treatment of IFN-alpha was safe and effective for the girl's clinical manifestations, and IFN-alpha might be a valuable first-line therapy for paediatric ECD.	23757907

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2013 May 6	Int J Cardiol	Erdheim-Chester disease with cardiac involvement successfully treated with anakinra	Killu AM, Liang JJ, Jaffe AS	Department of internal Medicine, Mayo Clinic, Rochester, MN, USA. Electronic address: killu.ammar@mayo.edu	No abstract available.	23659884
2013 May 1	Clin Nucl Med	FDG PET Images in a Patient With Erdheim-Chester Disease	Sioka, Chrissa MD; Estrada-Veras, Juvianee MD; Maric, Irinia MD; Gahl, William A. MD; Chen, Clara C. MD	Juvianee Estrada-Veras, MD, National Human Genome Research Institute, National Institutes of Health 10 Center Drive, Bldg 10, CRC- Room 3-2551 Bethesda, MD 20892-1851 juvianee.estradaveras@nih.gov	Erdheim-Chester disease is an uncommon non-Langerhans-cell histiocytosis, due to excessive production of histiocytes deposited in various organs and tissues in the human body. FDG PET was performed in a 68-year-old man with documented active Erdheim-Chester disease to evaluate the extent of the disease. The patient was previously treated with high-dose subcutaneous Interferon [alpha]2b, 1,000,000 units 3 times a week, but treatment was interrupted approximately 5 weeks before evaluation at the National Institutes of Health because of adverse effects of the medication. FDG PET/CT showed lesions were imaged in brain, heart, mediastinum, abdomen, and skeleton.	
2013 Apr 18	Clin Nucl Med	18F-Fluoride PET/CT Aspect of an Unusual Case of Erdheim-Chester Disease With Histologic Features of Langerhans Cell Histiocytosis	Caoduro, Cécile; Ungureanu, Constantin Marius; Rudenko, Boris; Angoue, Orland; Blagosklonov, Oleg; Paycha, Frédéric; Boulahdour, Hatem	Oleg Blagosklonov, Service de Médecine Nucléaire, CHU Jean Minjoz, 3 bv. Fleming, 25030 Besançon, France. E-mail: oleg.blagosklonov@univ-fcomte.fr	We report the case of a 63-year-old woman with Erdheim-Chester disease (ECD) and histologic features of Langerhans cell histiocytosis, both extremely rare histiocytic proliferations responsible of skeletal and extraskeletal involvement. F-Fluoride PET/CT revealed multiple intense focal uptake scattered throughout the skeleton. We also performed an F-FDG PET/CT which point out visceral and vascular involvement. This case illustrates the interest of PET/CT in ECD, a rare polymorphus and systemic disease, and in our knowledge, this is the first reported illustration of F-fluoride PET/CT findings in this pathology.	23603579
2013 May	Rheum Dis Clin North Am.	Erdheim-chester disease	Haroche J, Arnaud L, Cohen-Aubart F, Hervier B, Charlotte F, Emile JF, Amoura Z.	Department of Internal Medicine, French Reference Center for Rare Autoimmune and Systemic Diseases, Assistance Publique-Hôpitaux de Paris, Pitié-Salpêtrière Hospital, 47-83 boulevard de l'Hôpital, 75651 Paris Cedex 13, France; Université Pierre et Marie Curie, UPMC Univ Paris, Paris, France. Electronic address: julien.haroche@psl.aphp.fr	Erdheim-Chester disease (ECD) is a rare form of non-Langerhans' cell histiocytosis. Diagnosis of ECD is based on the identification in tissue biopsy of histiocytes, which are typically foamy and immunostain for CD68+ CD1a-. Central nervous system involvement is a major prognostic factor in ECD. Interferon alpha may be the best first-line therapy and significantly improves survival of ECD. The BRAFV600E mutation is found in more than 50% of cases. Vemurafenib has been used for a small number of patients harbouring this mutation; inhibition of BRAF activation by vemurafenib was highly beneficial in these cases of severe multisystemic and refractory ECD.	23597965

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Apr 15	BMJ Case Rep.	Erdheim-Chester disease with isolated craniocerebral involvement	Jain RS, Sannegowda RB, Jain R, Mathur T.	Department of Neurology, SMS Medical College Hospital, Jaipur, Rajasthan, India. drrsjain@yahoo.com, padayappa4u@yahoo.com	Erdheim-Chester disease (ECD) is a rare non-Langerhans form of histiocytosis with distinctive radiographic and pathological features. Intracranial involvement is further a rarity, usually producing diabetes insipidus or cerebellar-brainstem symptoms. We report a 40-year-old man presenting with recurrent secondarily generalised seizures. An MRI scan of the brain revealed multiple enhancing intracranial masses in frontal, temporal and parietal regions. Biopsy from the left frontotemporal lesion confirmed it to be a rare case of ECD. The patient received a short course of corticosteroids initially and subsequently remained well-controlled on antiepileptic therapy alone. A repeat MRI of his brain showed significant resolution of lesions. Osteolytic lesions in the skull vault were detected during follow-up which also disappeared. Interestingly, there was no involvement of long bones or any other system even after 12 years of follow-up.	23592809
2013 Apr 16	Ann Rheum Dis	Diagnosing Erdheim-Chester disease	Cavalli G, Berti A, Campochiaro C, Dagna L.	Unit of Medicine and Clinical Immunology, Vita-Salute San Raffaele University and San Raffaele Scientific Institute, Milan, Italy dagna.lorenzo@hsr.it	No abstract available.	23592711
2013 Mar 16	Ann Rheum Dis	Erdheim-Chester disease	Juanós-Iborra M, Solanich-Moreno J, Selva-O'Callaghan A.	Internal Medicine Department, Vall d'Hebron General Hospital, Universitat Autonoma de Barcelona, Barcelona, Spain	No abstract available.	23505236
2013 Mar	Clin Imaging	Suprasellar non-Langerhans cell histiocytosis (Erdheim-Chester disease)-a case report.	Sharma M, Vettiyil B, Bartlett E, Yu E	Head and Neck Imaging, Princess Margaret Hospital, Toronto M5G 2M9. Electronic address: drmanassharma@gmail.com	Erdheim-Chester disease (ECD) is an uncommon non-Langerhans cell histiocytosis that affects multiple body systems and can present clinically in a myriad of ways. An adult onset is most common with bony involvement and constitutional symptoms. We report the case of a 52-year-old female presenting with diabetes insipidus and a suprasellar mass on imaging, with no evidence of extracerebral involvement. Histopathology was consistent with ECD.	23465991
2013 Feb 26	Med Clin (Barc)	Erdheim-Chester disease: Report of a case with retroperitoneal involvement treated with interferon alpha-2a	Gil Ortega M, López Lozano E, Girela Baena E, Navarro Martinez N	Departamento de Medicina Interna, Hospital Morales Meseguer, Murcia, España. Electronic address: milagros.gilortega@gmail.com	Spanish. No abstract available.	23452672

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2013 Feb 28	Diagn Cytopathol	Erdheim-Chester Disease With Prominent Pericardial Effusion: Cytologic Findings and Review of the Literature	Alexiev BA, Staats PN	Borislav A. Alexiev, M.D., Department of Pathology, NBW85, University of Maryland Medical Center, 22 S Greene Street, Baltimore 21201, MD. E-mail: balexiev@umm.edu	Erdheim-Chester disease (ECD) is a rare, non-Langerhans form of histiocytosis of unknown origin with distinct clinicopathologic and radiographic features. Reports detailing the cytology of ECD are rare. We describe a case of ECD with pericardial effusion. Cytologic examination revealed a hypercellular specimen composed of clusters and singly dispersed foamy macrophages with round nuclei and inconspicuous nucleoli, admixed with lymphocytes, eosinophils, and Touton-type multinucleated giant cells. Immunostains for CD68 were strongly positive in the foamy macrophages while S100 and CD1a were negative. The presence of foamy histiocytes, multinucleated giant cells, lymphocytes and eosinophils are also features of other systemic histiocytic disorders, including Langerhans cell histiocytosis (LCH), Rosai-Dorfman disease (RDD) and sarcoidosis. To the best of out knowledge, this is the first report describing the cytological features of ECD in a pericardial effusion.	3447119
2013 Feb 19	Diagn Interv Imaging	Erdheim-Chester disease: A rare diagnosis with evocative imaging	Alberti N, Frulio N, Bertolotti A, Petitpierre F, Veron A, Perez JT, Raffray L, Vanquaetem H, Salut C, Pinaquy JB, Trillaud H	Service de radiologie, hôpital Saint-André, 1, rue Burguet, 33075 Bordeaux, France. Electronic address: nicolasalberti@aol.com	No abstract available	23433542
2013 Feb 8	Ann Rheum Dis.	The multifaceted clinical presentations and manifestations of Erdheim-Chester disease: comprehensive review of the literature and of 10 new cases.	Cavalli G, Guglielmi B, Berti A, Campochiaro C, Sabbadini MG, Dagna L lorenzo.dagna@unisr.it	1 Vita-Salute San Raffaele University, Milan, Italy	OBJECTIVES: Erdheim-Chester disease (ECD) is a rare inflammatory disorder characterised by organ infiltration by non-Langerhans' histiocytes. Although rare, ECD is clearly an overlooked diagnosis. No data specifically addressing the most frequent presentations of ECD at the time of onset in a large cohort of patients are currently available. METHODS: We reviewed all the published cases in the English literature of histologically-confirmed ECD. We excluded reports in which data regarding onset and diagnosis were not univocal, as well as repeated reports of the same case(s). We also included in the analysis 10 new unpublished patients from our cohort. We analysed the disease presentation with particular regard to the manifestations that induced patients to seek medical attention and their subsequent evolution. RESULTS: In the cumulative cohort of 259 cases, ECD predominantly presented with skeletal symptoms, diabetes insipidus, neurological and constitutional symptoms. Diabetes insipidus and constitutional symptoms, if not present at onset, seemed to only seldom develop. There were differences in ECD presentation and course among different age groups of patients. CONCLUSIONS: Physicians should be aware of the extraordinarily heterogeneous clinical presentations and manifestations of ECD in order to include ECD in the differential diagnosis of several conditions.	23396641
2013 Jan 26	Cerebellum	A Rare Cause of Late-Onset Cerebellar Ataxia: Erdheim-Chester Disease	Shanmugam SV, Kolappan M, Garg M, Rennie WJ, Furness P, Rajabally YA	Department of Neurology, University Hospitals of Leicester, Leicester, LE5 4PW, UK y.rajabally@btinternet.com	No abstract available	23354540

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012	Duodecim	Thirst and extensive bone lesions in a previously basically healthy woman	Laitinen K, Lamminen A, Anttila P, Lohman M, Karjalainen-Lindsberg ML.	kalevi.laitinen@hus.fi	A previously quite healthy 65-year-old woman sought emergency hospital care due to fatigue, weight loss and sensation of thirst appearing over a couple of months. Further analysis revealed a process affecting the neurohypophysis and extensive lytic sclerotic bone lesions. Eventually a rare generalized underlying disease was unraveled: the diagnosis included both Langerhans cell histiocytosis and Erdheim-Chester disease.	23342482
2012 Dec	Cancer Res Treat.	Reply to commentary on "a case of erdheim-chester disease with asymptomatic renal involvement"	Lee HJ, Kim TM.	Tae Min Kim, MD. Department of Internal Medicine, Seoul National University Hospital, Seoul National University College of Medicine, 101 Daehak-ro, Jongno-gu, Seoul 110-744, Korea. Tel: +82-2-2072-3559, Fax: +82-2-764-2199, Email: gabriel9@snu.ac.kr	No abstract available	23341795
2012 Dec	Cancer Res Treat.	Commentary on "a case of erdheim-chester disease with asymptomatic renal involvement"	Li Cavoli G.	Division of Nephrology and Dialysis, Civic and Di Cristina Hospital, Gioacchino Li Cavoli, via Francesco Cilea 43, Palermo 90144, Italy. Tel: 39-3332318100, Fax: 39-0916663454, Email: gioacchinolicavoli@libero.it	No abstract available	23341794
2013 Jan 8	Oncologist	Hand-Schuller-Christian Disease and Erdheim-Chester Disease: Coexistence and Discrepancy	Yin J, Zhang F, Zhang H, Shen L, Li Q, Hu S, Tian Q, Bao Y, Jia W.	Weiping Jia, M.D., Ph.D., Shanghai Clinical Center for Diabetes, Department of Endocrinology and Metabolism, Shanghai Jiao Tong University Affiliated Sixth People's Hospital, 600 Yishan Road, Shanghai 200233, China. Telephone: 011-86-21-64369181; Fax: 011-86-21-64368011; E-mail: wpjia@sjtu.edu.cn	Langerhans cell histiocytosis (LCH) and Erdheim-Chester disease (ECD) share similar clinical features and mechanisms. In very rare circumstances, the two diseases coexist in the same patient. Here we report such a patient, who was first diagnosed with Hand-Schüller-Christian disease (HSC), a type of LCH. Several years later, the patient presented with severe exophthalmos and osteosclerosis on radiograph. New biopsy revealed ECD. We also analyze 54 cases of LCH and 6 cases of ECD diagnosed in our hospital, as well as their progression during a follow-up period of 8 years. In five cases of HSC (9.3% of LCH), a triad of central diabetes insipidus, hyperprolactinemia, and pituitary stalk thickening on magnetic resonance imaging (MRI) preceded the typical bone lesions by 4-9 years. In addition, LCH was featured as elevated plasma alkaline phosphatase (ALP), which was normal in ECD. Combined with a literature review, several features are summarized to differentiate ECD from HSC. In patients with diabetes insipidus, concomitant hyperprolactinemia and pituitary stalk thickening on MRI indicate a possible HSC. Additionally, if osteosclerosis is observed in a patient with LCH, the coexistence of ECD should be considered.	23299772

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Dec 20	Blood	Dramatic efficacy of vemurafenib in both multisystemic and refractory Erdheim-Chester disease and Langerhans cell histiocytosis harboring the BRAF V600E mutation	Haroche J, Cohen-Aubart F, Emile JF, Arnaud L, Maksud P, Charlotte F, Cluzel P, Drier A, Hervier B, Benameur N, Besnard S, Donadieu J, Amoura Z.	Department of internal medicine & French reference center for rare auto-immune & systemic diseases, AP-HP, Pitie-Salpetriere hospital, Paris, France julien.haroche@psl.aphp.fr	Histiocytoses are rare disorders of unknown origin with highly heterogeneous prognosis. BRAF(V600E) gain-of-function mutations have been observed in 57% of cases of Langerhans cell histiocytosis (LCH) and 54% of cases of Erdheim-Chester disease (ECD), but not in other types of histiocytoses. Targeted therapy with an inhibitor of mutated BRAF (vemurafenib) improves survival of patients with melanoma. Here, we report vemurafenib treatment of three patients with multisystemic and refractory ECD carrying the BRAF(V600E) mutation; two also had skin or lymph node LCH involvement. The patients were assessed clinically, biologically (CRP values), histologically (skin biopsy), and morphologically (positron emission tomography [PET], computed tomography and magnetic resonance imaging). For all patients, vemurafenib treatment led to substantial and rapid clinical and biological improvement, and the tumor response was confirmed by PET, computed tomography and/or magnetic resonance imaging one month after treatment initiation. For the first patient treated, the PET response increased between months one and four of treatment. The treatment remained effective after 4 months of follow-up although persistent disease activity was still observed. Treatment with vemurafenib, a newly approved BRAF inhibitor, should be considered for patients with severe and refractory BRAF(V600E) histiocytoses, particularly when the disease is life-threatening.	23258922

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Nov	Vnitr Lek	The effect of lenalidomide on rare blood disorders: Langerhans cell histiocytosis, multicentric Castleman disease, POEMS syndrome, Erdheim-Chester disease and angiomatosis	Adam Z, Pour L, Krejčí M, Zahradová L, Szturz P, Koukalová R, Rehák Z, Nebeský T, Hájek R, Král Z, Mayer J.	Interní hematoonkologická klinika, LF MU a FN Brno. z.adam@fnbrno.cz	Lenalidomide has been licenced for the treatment of multiple myeloma and, in 2012, it is used as a standard treatment of relapses of the disease. Literature contains a number of publications on the effects of lenalidomide in myelodysplastic syndrome, in malignant lymphomas and chronic B lymphocytic leukaemia. The effects of the drug in rare diseases, however, have not been investigated so far. In this paper, we summarize our experience with lenalidomide in rare blood disorders. We observed an excellent effect of lenalidomide in multifocal aggressive, repeatedly relapsing Langerhans cell histiocytosis where it led to complete remission. This patient was treated with 2-chlorodeoxyadenosine and with CHOEP (cyclophosphamide, etoposide, doxorubicin, vincristine and prednisone) chemotherapy and high dose BEAM chemotherapy with autologous transplantation of haematopoietic tissue for an early disease relapse. Following another early relapse, the patient was treated with lenalidomide (25 mg). Treatment with lenalidomide induced complete remission on PET-CT. The patient was consolidated during the remission with a reduced intensity conditioning regimen and allogeneic transplantation of haematopoietic tissue. Following allogeneic transplantation, the patient has been in full remission for 10 months. We further showed an excellent effect of lenalidomide in multicentric Castleman disease with generalized involvement of lymphatic nodes, B symptoms and vasculitis. We observed partial efficacy in Erdheim-Chester disease. We used 2-chlorodeoxyadenosine as part of initial treatment that delivered partial regression of brain infiltrates only; fluorodeoxyglucose accumulation in the bones has not changed. Lenalidomide 25 mg was used as second line treatment. This led to complete regression of CNS infiltrates on MRI but fluorodeoxyglucose accumulation in bone lesions did not change. Regression of clinical signs and regression of fibrosis of retroperitoneum was achieved with an ongoing treatment with anakinra. Conclusion: We showed an effect of lenalidomide in Langerhans cell histiocytosis and in Castleman disease. The treatment led to regression of brain infiltrates in a patient with Erdheim-Chester disease. A dose of 10 mg of lenalidomide daily in combination with 50 mg of thalidomide stabilized a course of angiomatosis. Lenalidomide did not deliver the required treatment response in a patient with POEMS syndrome and multiple previous therapies. [Publised in Czech.]	23256832
2012 Dec 17	J Clin Oncol	BRAF Mutations in Erdheim-Chester Disease.	Emile JF, Charlotte F, Amoura Z, Haroche J.	Department of internal medicine & French reference center for rare auto-immune & systemic diseases, AP-HP, Pitie-Salpetriere hospital, Paris, France julien.haroche@psl.aphp.fr	No abstract available	23248255

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Oct 11	Hum Pathol	Histiocytic disorders of the gastrointestinal tract	Detlefsen S, Fagerberg CR, Ousager LB, Lindebjerg J, Marcussen N, Nathan T, Sørensen FB	Department of Clinical Pathology, Vejle Hospital, 7100 Vejle, Denmark; Department of Pathology, Odense University Hospital, 5000 Odense, Denmark. Electronic address: S.Detlefsen@gmx.net.	The morphologic diagnosis of histiocytic lesions of the gastrointestinal tract can be challenging, and several disorders have to be considered in their differential diagnosis. We present one of the most widespread examples of xanthomatosis of the gastrointestinal tract published so far and give a short review on histiocytic disorders of the gastrointestinal tract in general. The primary histiocytic disorders of uncertain origin, Rosai-Dorfman disease, Langerhans cell histiocytosis, and Erdheim-Chester disease, are addressed. Reactive and infectious conditions such as xanthomatosis, xanthogranulomatous inflammation, juvenile xanthogranuloma, Whipple's disease and malacoplakia are discussed as well. We also briefly go into primary histiocytic disorders of neoplastic origin, systemic diseases with secondary gastrointestinal tract involvement like the lysosomal storage disorders, and pigmented lesions. Using a panel of histochemical stains and immunohistochemical markers, together with conventional microscopy, clinical information, and imaging studies, the diagnosis of histiocytic disorders of the gastrointestinal tract can be established in most instances.	23063502
2012 Oct 1.	Intern Med	Immunopathological analysis of erdheim-chester disease with massive ascites	Ota M, Sakamoto M, Sato K, Yoshida Y, Funakubo Asanuma Y, Akiyama Y, Yamakawa M, Mimura T.	Department of Rheumatology and Applied Immunology, Faculty of Medicine, Saitama Medical University, Japan 2) Department of Pathological Diagnostics, Faculty of Medicine, Yamagata University, Japan Correspondence to Dr. Kojiro Sato, satok@saitama-med.ac.jp	We treated a 77-year-old woman with pleural and pericardial effusion and ascites. Initially, collagen vascular disease was suspected due to the presence of anti-centromere antibodies and suspected complication of pulmonary arterial hypertension. However, soft-tissue abnormalities surrounding the bilateral kidneys detected on computed tomography (CT) and symmetrical lesions of the long bones detected on bone scintigraphy made us consider a diagnosis of Erdheim-Chester disease (ECD), which is a rare form of histiocytosis. We immunochemically analyzed the cells derived from the ascites in detail and confirmed the diagnosis. Immunocytochemical analyses may therefore help to achieve a better understanding of the pathogenesis of this rare disease.	23037484
2012 Jul-Aug	JBR-BTR	Erdheim-Chester disease detected with 99mTc MDP bone SPECT/CT	Ceulemans G, Keyaerts M, Verbruggen L, Hoorens A, Boulet C, Verdries D, De Maeseneer M, Ilsen B, Everaert H.	Department of Nuclear Medicine, UZ Brussel, Brussels, Belgium	Erdheim-Chester disease (ECD) is a rare non-Langerhans' cell histiocytosis. Mild but permanent juxta-articular bone pain in mainly knees and ankles is the most frequent associated symptom. Despite the pathognomonic radiographic findings, most cases are still diagnosed by the pathologist.The lesions consist of lipid-storing CD 68 +/CD 1a--non-Langerhans' cell histiocytes, most frequently localized in bone but also involving multiple organ systems in the body. We present a case report in which the diagnosis of ECD was established with 99mTc MDP bone SPECT/CT.	23019992
2012 Sep 24	J Clin Oncol	Erdheim-Chester Disease Harboring the BRAF V600E Mutation	Blombery P, Wong SQ, Lade S, Prince HM.	Peter MacCallum Cancer Centre, Victoria, Australia blombery@bigpond.com	No abstract available.	23008323

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Sep 19	Joint Bone Spine	Favorable radiological outcome of skeletal Erdheim-Chester disease involvement with anakinra	Aubert O, Aouba A, Deshayes S, Georgin-Lavialle S, Rieu P, Hermine O.	Department of Adult Haematology, Centre National de la Recherche Scientifique, Unité Mixte de Recherche 8147, Hôpital-Necker Enfants-Malades, Université de Paris-Descartes, AP-HP, Paris, France. doc.aubert@gmail.com	Erdheim-Chester disease is a rare non-langerhans cell histiocytosis characterized by infiltration of foamy CD68-positive but CD1a-negative macrophages and fibro-inflammatory lesions as retroperitoneal, periureteral areas or bones. Interferon-α therapy has been used as treatment but it had variable efficiency and limited tolerance. More recently, a recombinant form of interleukin-1 receptor antagonist (anakinra) was used with success but no skeletal radiological improvement was recorded. We report here a case of interleukin-1 receptor antagonist in the treatment of refractory bones infiltration in Erdheim-Chester disease. After 1 year of treatment, the positron emission tomography-computed tomography showed an outstanding response of the skeletal involvement with clearly lower and smaller hypermetabolism images.	22999905
2012 Sep 11	Clin Neurol Neurosurg.	Paroxysmal exertion-induced dystonia secondary to Erdheim-Chester disease.	Baldacci F, Lucetti C, Vergallo A, Borelli P, Tessa C, Viacava P, Bonuccelli U.	Department of Neuroscience, University of Pisa, Pisa, Italy. f.baldacci79@yahoo.it.	No abstract available.	22980524
2012 Oct;12	Pract Neurol.	Histiocytosis for the neurologist: a case of Erdheim-Chester disease.	Selvarajah JR, Rodrigues MG, Ali S.	Dr Johann R Selvarajah, Institute of Neurological Sciences, Southern General Hospital, Glasgow, UK; Johann.Selvarajah@ggc.scot.nhs.uk	The histiocytoses are a rare but diverse group of disorders, ranging from localised, self-limiting lesions to disseminated, fulminant, multi-system disease. Some histiocytoses may cause or present with neurological disease and their recognition can be challenging. We illustrate this with a case, followed by a discussion of the clinical characteristics and management of the more common histiocytoses that may present to the neurologist.	22976062
2012 Aug	Arch Neurol.	Erdheim-Chester Disease	Liotta EM, Jhaveri MD, Fox JC, Venugopal P, Lewis SL.	Steven L. Lewis, MD Dept. of Neurological Sciences Rush University Medical Center 1725 W. Harrison St., Ste. 1106 Chicago, IL 60612 slewis@rush.edu	No abstract available.	22925950
2012 Jun;14	Isr Med Assoc J.	Erdhiem-Chester disease in a 49 year old man.	Asher I, Rabinovith I, Katz M, Sthoeger Z.	Dr. Z. Sthoeger Head, Depts. of Internal Medicine B and Allergy, Clinical Immunology and Neve-Or AIDS Center, Kaplan Medical Center, Rehovot 76100, Israel Phone: (972-8) 944-1403 Fax: (972-8) 944-1826 email: Zev_s@clalit.org.il	No abstract available.	22891408

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Jun	Vnitr Lek	Cladribine is highly effective in the treatment of Langerhans cell histiocytosis and rare histiocytic disorders of the juvenile xanthogranuloma group	Adam Z, Szturz P, Pour L, Krejčí M, Zahradová L, Tomíška M, Král Z, Koukalová R, Rehák Z, Mayer J	Interní hematoonkologická klinika, LF MU a FN Brno. z.adam@fnbrno.cz	Cladribine (2-chlorodeoxyadenosine) is metabolised and phosphorylated in a cell up to 2-chloroadenosine triphosphate which is the actual effective form of the drug. The greatest accumulation of 2-chloroadenosine triphosphate is in the most active cells, where activating (phosphorylation) enzyme, deoxycytidine kinase, has the highest activity, whereas inactivating enzyme (dephosphorylation), cytoplasmic 5-nucleotidase, has the lowest activity. A very good ratio of the both enzymes for high effectiveness of cladribine is in resting and proliferating lymphocytes. Therefore, cladribine is an effective medication for hairy cell leukemia, Waldenström macroglo-bulinemia but also for chronic -B-lymphocytic leukemia. However, such high concentrations of 2-chloroadenosine triphosphate are reached in some cells of histiocytic lines, in monocytes and also in Langerhans dendritic cells. Thats why cladribine is highly effective medication in treating Langerhans cell histiocytosis and also in treating diseases of the juvenile xanthogranuloma group. In the paper we present a survey of published experience with cladribine in patients with Langerhans cell histiocytosis. The effectiveness of cladribine in the childhood form of Langerhans cell histiocytosis is investigated only in 1 multicentric clinical study, other data are taken from single case reports or small series studies. Cladribine was used in 60 adult patients altogether and in 51 of them (85%) treatment response (CR + PR) was achieved. In the group of childhood patients cladribine was used in 182 cases and treatment response (CR + PR) was reached in 110 (60.4%) thereof. One possible explanation for a higher number of therapy responses in adults is lower Langerhans cell histiocytosis aggressiveness in adults than in children. Another explanation is the fact that therapy responses in adults are summarized only from case reports and smaller cohorts, whereas in children, case reports and also results of a prospective randomized clinical study are included. Diseases of the juvenile xanthogranuloma group are much more rare than Langerhans cell histiocytosis and so the number of publications is smaller. In total, 7 publications describe therapy response of cladribine in some of the juvenile xanthogranuloma forms (Erdheim-Chester disease, disseminated juvenile xanthogranuloma and localized form of plane xanthoma type). Cladribine was also effective in CNS infiltration by Langerhans cell histiocytosis cells or juvenile xanthogranuloma cells. Conclusions: Cladribine is a highly effective medication used in treating Langerhans cell histiocytosis. It is very good tolerated in monotherapy. Therefore, it is suitable for initial therapy of adults with multifocal or multisystem form of Langerhans cell histiocytosis. Furthermore, it has the use in treating relapses after some other initial therapy. According to published experience, it is an effective drug for diseases of the juvenile xanthogranuloma group (Erdheim-Chester disease, diffuse juvenile xanthogranuloma and also Rosai-Dorfman disease). Key words: cladribine - 2-chlorodeoxyadenosine - Langerhans cell histiocytosis - diabetes insipidus - juvenile xanthogranuloma - plane xanthoma - Erdheim-Chester disease - Rosai-Dorfman disease - sinus histiocytosis with massive lymphadenopathy. [Article in Czech]	22913238

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Jun;14	Isr Med Assoc J	Erdheim-Chester disease: an orphan condition seeking treatment	Mazor RD, Kesler A, Shoenfeld Y.	Dr. Y. Shoenfeld Zabludowicz Center for Autoimmune Diseases, Sheba Medical Center, Tel Hashomer 52621, Israel Phone: (972-3) 530-8070, Fax: (972-3) 535-2855 email: shoenfel@post.tau.ac.il	Comment on Erdhiem-Chester disease in a 49 year old man.	22891403
2012	Malaysian Orthopaedic Journal	Rare Presentation of a Rare Disease (Erdheim-Chester disease): A Case Report	Koh TW, M Fadli, SL Vijaya Kumar, Ashutosh S Rao	SL Vijaya Kumar, Department of Orthopaedics, Sultan Abdul Halim Hospital, 08000 Sungai Petani, Malaysia Email: vksuppan@hotmail.com	Erdheim-Chester disease (ECD) was first reported by J. Erdheim and W. Chester, in 1930. There are less than 250 reported cases till date. We report a case of ECD in a 16- year-old Malay male, who initially presented with elusive anemic symptoms with more specific symptoms of bony pain, cardiorespiratory and hepatic involvement evolving as the disease progressed.	
2012 August	Open Journal of Rheumatology and Autoimmune Diseases	Erdheim-Chester Disease with Right Atrial Tumor and “Temporal Arteritis”	Joseph Skalski, William D. Edwards, Eric L. Matteson	Division of Rheumatology, Mayo Clinic College of Medicine, Rochester, USA. Email: matteson.eric@mayo.edu	Erdheim-Chester disease is an unusual syndrome characterized by non-Langerhans cell histiocytosis which can mimic rheumatologic diseases. We report a case of Erdheim-Chester masquerading as giant cell arteritis, which was success-fully managed with anakinra as anti IL-1 therapy.	
2012 Aug 9	Blood	High prevalence of BRAF V600E mutations in Erdheim-Chester disease but not in other non-Langerhans cell histiocytoses	Haroche J, Charlotte F, Arnaud L, Von Deimling A, Hélias-Rodzewicz Z, Hervier B, Cohen-Aubart F, Launay D, Lesot A, Mokhtari K, Canioni D, Galmiche L, Rose C, Schmalzing M, Croockewit S, Kambouchner M, Copin MC, Fraitag S, Sahm F, Brousse N, Amoura Z, Donadieu J, Emile JF	Department of internal medicine & French reference center for rare auto-immune & systemic diseases, AP-HP, Pitie-Salpetriere hospital, Paris, France julien.haroche@psl.aphp.fr	Histiocytoses are rare disorders of unknown origin with highly heterogeneous prognosis. BRAF mutations have been observed in Langerhans cell histiocytosis (LCH). We investigated the frequency of BRAF mutations in several types of histiocytoses. Histology from 127 patients with histiocytoseswere reviewed. Detection of BRAF(V600) mutations was performed by pyrosequencing of DNA extracted from paraffin embedded samples. Diagnosis of Erdheim-Chester Disease (ECD), LCH, Rosai-Dorfman disease, juvenile xanthogranuloma, histiocytic sarcoma, xanthoma disseminatum, interdigitating dendritic cell sarcoma and necrobiotic xanthogranuloma were performed in 46, 39, 23, 12, 3, 2, 1 and 1 patients respectively. BRAF status was obtained in 93 cases. BRAF(V600) mutations were detected in 13/24 (54%) ECD, 11/29 (38%) LCH, and none of the other histiocytoses. Four patients with ECD died of disease. The high frequency of BRAF(V600) in LCH and ECD suggests a common origin of these diseases. Treatment with vemurafenib should be investigated in patients with malignant BRAF(V600) histiocytosis.	22879539

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Aug 6	J Clin Oncol	Tumor Necrosis Factor α As a Master Regulator of Inflammation in Erdheim-Chester Disease: Rationale for the Treatment of Patients With Infliximab	Dagna L, Corti A, Langheim S, Guglielmi B, De Cobelli F, Doglioni C, Fragasso G, Sabbadini MG, Ferrarini M.	Vita-Salute San Raffaele University; San Raffaele Scientific Institute, Milan, Italy. dagna.lorenzo@hsr.it	No abstract available. (Supported in part by the ECD Global Alliance, DeRidder, LA., with final grant report available. E-mail: support@erdheim-chester.org.)	22869874
2012 Aug 4	Clin Exp Nephrol	Image of Erdheim-Chester disease requiring hemodialysis	Tsukamoto M, Akahane M, Nangaku M.	Department of Nephrology and Endocrinology, Graduate School of Medicine, The University of Tokyo, 7-3-1 Hongo, Bunkyo-ku, Tokyo, 113-8655, Japan.	No abstract available.	22864518
2012 May	J Heart Valve Dis	Postoperative cardiac homograft involvement in Erdheim-Chester disease	Siram AT, Kouvatsos T, Suarez Y, Wohler A, Stelzer P, Strauchen J, Mechanick JI	Division of Endocrinology, Diabetes and Bone Diseases, Mount Sinai Medical Center, New York, NY 10029, USA. amulyasiram@gmail.com	Erdheim-Chester disease (ECD) is a rare multisystem disorder which is known to affect the skin, lungs, bone, pituitary gland, retroperitoneum and cardiovascular system. The case is described of a patient with ECD who had previously undergone a Ross procedure for presumed endocarditis involving the aortic valve and aortic root. The patient subsequently developed arthralgias, abdominal pain (requiring an exploratory laparotomy) and polydipsia. Furthermore, he developed progressive, symptomatic stenosis of the pulmonic homograft. A reoperative replacement of the homograft was required. The clinically suspected diagnosis of ECD was confirmed by a pathologic analysis of the explanted pulmonary homograft, and also (retrospectively) of previously resected mesenteric tissue. It is postulated that the patient may have developed ECD as a result of an immunologic reaction to the homograft tissue used for the Ross procedure.	22808847
2012 Jun	Cancer Res Treat	A case of erdheim-chester disease with asymptomatic renal involvement	Lee HJ, Lee KY, Shin DY, Lee YG, Choi SY, Moon KC, Han IK, Kim TM	Department of Internal Medicine, Seoul National University Hospital, Seoul National University College of Medicine, Seoul, Korea. gabriel9@snu.ac.kr	Erdheim-Chester disease is a rare non-Langerhans-cell histiocytosis involving bones and multiple organs. Its clinical course can vary, from an asymptomatic state to a fatal disease, with renal involvement being a common cause of death. A 41-year-old man presented with a 10-month history of bilateral lower limb pain. Left perirenal soft-tissue infiltration had been found incidentally two years earlier. No progression of the lesion or deterioration of renal function was observed for a period of two years. At admission, plain radiography and magnetic resonance imaging of the patient's lower limbs showed patchy osteosclerosis. Biopsy of the tibia revealed histiocytic infiltration, which was found to be positive for CD68 and negative for CD1a. This report describes an unusual case of Erdheim-Chester disease involving a stationary course of disease with no specific treatment for a long period of time.	22802754

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Jul 11	Med Clin (Barc)	Erdheim-Chester disease: study of 12 cases	Juanós Iborra M, Selva-O'Callaghan A, Solanich Moreno J, Vidaller-Palacin A, Martí S, Grau Junyent JM, Vilardell Tarrés M	Servicio de Medicina Interna, Hospital General Universitario Vall d'Hebron, Departamento de Medicina, Universitat Autònoma de Barcelona, Barcelona, España. aselva@vhebron.net	BACKGROUND AND OBJECTIVE: Erdheim-Chester disease (EC) is a rare form of non-Langerhans' cell histiocytosis. It is characterized by the xanthomatous infiltration of tissues with foamy CD68+/CD1a- histiocytes. We report a series of 12 patients diagnosed with EC. PATIENTS AND METHODS: We reviewed the clinical, pathological and therapeutic aspects of 12 cases diagnosed with EC at 7 tertiary teaching hospitals in Spain. Patients were included if tissue infiltration by histiocytes CD68+/CD1a- could be demonstrated in an appropriate clinical setting. RESULTS: Twelve patients (7 male) were included. Median follow-up was 36 months (IQR: 20-84). The median age at the time of clinical onset and pathological diagnosis was 49 (IQR: 28-61) and 56 years (IQR: 37-62), respectively. In 6 cases multiples biopsies were performed (skin, muscle, testicular) previous to diagnosis, which was confirmed in 3 cases after a carefully review of pathological tissues. Neurological involvement was independently associated with mortality (P<.05). Characteristic long bone osteosclerosis was detected in 9 patients. CONCLUSION: EC is a multisystemic and heterogeneous clinicopathological condition. A high index of suspicion and fluent communication between clinicians and pathologists is necessary to achieve a correct diagnosis. [Article in Spanish]	22795496
2012 Jul	Radiographics	Tumorlike conditions of the pleura	Walker CM, Takasugi JE, Chung JH, Reddy GP, Done SL, Pipavath SN, Schmidt RA, Godwin JD 2nd	Departments of Radiology and Pathology, University of Washington Medical Center, 1959 NE Pacific St, Box 357115, Seattle, WA 98195; Puget-Sound VA Health Care System and Seattle Children's Hospital, Seattle, Wash. walk0060@uw.edu	Tumorlike conditions of the pleura are rare, but diagnosis is facilitated by recognizing certain imaging patterns and interpreting them in the clinical context. A tumorlike condition of the pleura is any nonneoplastic lesion of the pleura itself, or within the pleural space, that resembles a tumor. An approach to diagnosis of the tumorlike conditions of the pleura is provided, and these conditions are grouped into focal or diffuse conditions, with an emphasis on specific imaging features. Focal tumorlike conditions of the pleura include pleural plaque, thoracic splenosis, thoracic endometriosis causing catamenial pneumothorax, and pseudotumor caused by pleural effusion. Thoracic splenosis should be considered in a patient who has a healed left lower rib fracture, an absent spleen, and left lower pleural nodules. Thoracic endometriosis with catamenial pneumothorax should be considered in a woman of childbearing age who presents with right scapular pain and recurrent pneumothorax occurring at or around the onset of menses. Extrapleural hematoma is a nonpleural mimic of pleural tumor and shares some imaging features with focal tumorlike conditions of the pleura, despite residing in the extrapleural space. Diffuse tumorlike conditions of the pleura include diffuse pleural thickening and rare conditions such as Erdheim-Chester disease and diffuse pulmonary lymphangiomatosis. Erdheim-Chester disease should be considered when diffuse pleural thickening occurs with a perirenal soft-tissue halo or distal femoral sclerosis. Diffuse pulmonary lymphangiomatosis should be considered when findings include diffuse pleural thickening, interlobular septal and peribronchovascular interstitial thickening, and mediastinal fat infiltration limited to the thorax and when these findings persist despite diuretic therapy.	22786988

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Jul;	Clin Nephrol	Pericardial effusion as a crucial presentation of Erdheim-Chester disease in a hemodialysis patient: an overlooked diagnosis	Chen MT, Wang SM, Lin SY, Ting IW, Liu JH, Kuo HL, Huang CC.	Division of Nephrology and Kidney Institute, Department of Internal Medicine, and China Medical University, Taichung, Taiwan. cch@www.cmuh.org.tw	Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis whose hallmark is tissue infiltration by CD68-positive, CD1a-negative and usually S-100 protein-positive foamy non-Langerhans histiocytes and mononuclear cells. Here, we report a hemodialysis (HD) patient who presented with fever and pericardial effusion. We performed pericardiocentesis with pericardial biopsy and the histological findings indicated ECD. We administered intravenous methylprednisolone pulse therapy (250 mg/d) followed by oral prednisolone (50 mg/d). The patient's fever gradually subsided and there was no recurrence of pericardial effusion. This is the first report of an HD patient with ECD. We suggest that ECD be considered in the differential diagnosis of new HD patients who present with pericardial effusion, especially when this did not improve following increased dose of HD.	22732342
2012 Jun 7	J Clin Neurosci	Xanthomatous hypophysitis	Niyazoglu M, Celik O, Bakkaloglu DV, Oz B, Tanriöver N, Gazioglu N, Kadioglu P.	Division of Endocrinology and Metabolism, Department of Internal Medicine, University of Istanbul, Cerrahpasa Medical School, Endokrinoloji-Metabolizma ve Diyabet Bilim Dali, Cerrahpasa 34303, Istanbul, Turkey. kadioglup@yahoo.com	Xanthomatous hypophysitis (XH) is the rarely seen primary form of hypophysitis. The histological differential diagnosis includes other causes of hypophysitis, Erdheim-Chester disease (ECD), Langerhans cell histiocytosis, Rosai-Dorfman disease and plasma cell granulomas. We present a 39-year-old woman admitted to our department with headache, menstrual irregularity and galactorrhea. The MRI revealed a lesion with a central cystic/necrotic region and a diameter of almost 1cm. Histologic examination showed an inflammatory infiltrate of numerous foamy histiocytes, surrounding the necrotic tissue. On immunohistochemical sections, infiltrating foamy cells stained strongly positive for CD68, and negative for CD1a and S100. After establishing the diagnosis of XH, the patient underwent glucocorticoid treatment. XH should be considered in the differential diagnosis of pituitary lesions. Since XH is rare, it is difficult to assess the efficacy of medical/surgical treatment of this entity accurately.	22682648
2012 Jun 3	Radiologe	Obscure exophthalmus	Götz A, Stroszczynski C, Müller-Wille R.	Institut für Röntgendiagnostik, Universitätsklinikum Regensburg, Franz-Josef-Strauß-Allee 11, 93053, Regensburg, Deutschland, andrea.goetz@ukr.de.	A 63-year-old male patient presented with retrobulbar pressure which had been present for 1 year. Contrast-enhanced computed tomography (CT) and magnetic resonance imaging (MRI) revealed symmetrical retrobulbar, perivascular and retroperitoneal infiltration of soft tissue and also showed cardiac involvement. In combination with the histological findings displaying infiltration by foamy histiocytes, Erdheim-Chester disease was diagnosed. [article in German.]	22660510
2012 May 26	J Neurol	Erdheim-Chester disease presenting with an intramedullary spinal cord lesion.	Tzoulis C, Gjerde IO, Søfteland E, Neckelmann G, Strøm E, Vintermyr OK, Sviland L, Biermann M.	Department of Neurology, Haukeland University Hospital, 5021, Bergen, Norway, chtzoulis@yahoo.com	--	22638564
2012 May 25	Orbit	Unusual Orbital Involvement in Erdheim Chester Disease: A Radiological Diagnosis	Arora A, Sharma S, Pushker N, Kashyap S, Bakhshi S.	Sanjay Sharma, AIIMS, Radiodiagnosis, Ansari Nagar, New Delhi, 110029 India. E-mail: drssharma@hotmail.com	Erdheim-Chester disease (ECD) is an exceedingly rare, disseminated non-Langerhan cell histiocytosis with multisystem involvement, having characteristic sclerotic skeletal lesions. We present an unusual case primarily manifesting as an extensive orbital disease, with low-grade systemic involvement. Owing to its rarity and therefore lack of general awareness it remains a difficult clinical and pathologic diagnosis. Immuno-histochemistry of the biopsy specimen is diagnostic.	22631449

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 May 21	Clin Radiol.	Neoplastic and proliferative disorders of the perinephric space	Heller MT, Haarer KA, Thomas E, Thaete FL.	Department of Radiology, University of Pittsburgh Medical Center, Pittsburgh, PA, USA. hellermt@upmc.edu	The perinephric space is a well-marginated central compartment of the retroperitoneum, located between the anterior and posterior pararenal spaces. Various neoplastic and proliferative disorders can affect the perinephric space, and there is a wide array of imaging findings. Although many perinephric lesions may extend directly from the kidney and adrenal gland, other lesions occur in the perinephric space due to haematogenous spread, as part of a systemic disease, or by extension from an adjacent retroperitoneal compartment. Imaging plays a pivotal role in the diagnosis of perinephric diseases, as many of the disease processes affecting this space will not result in clinical signs or symptoms until the disease is at an advanced stage. Despite the often shared non-specific clinical and imaging findings among these disease processes, application of a categorical differential diagnosis based on the imaging characteristics will serve to narrow the differential diagnosis and direct further evaluation and treatment. In this article, the lesions have been categorized as soft-tissue rind [nephroblastomatosis, fibrosis, Erdheim-Chester disease (ECD), extramedullary haematopoiesis, lymphoma, infiltrating metastases], focal solid lesions (extension of renal or adrenal malignancies, melanoma metastases, treated lymphoma), fat-containing lesions (angiomyolipoma, liposarcoma, myelolipoma), and cystic lesions (lymphangiomas, abscesses). The aim of this article is to demonstrate and describe the key imaging features of several neoplastic and proliferative disorders that affect the perinephric space.	22622354
2012 Jan-Feb	Indian Heart J.;	Recurrent and rapidly occurring pericardial tamponade in Erdheim Chester disease.	George OK, Subhendu MS.	Professor, Department of Cardiology, Christian Medical College, Vellore, India drsubhendu@gmail.com	Erdheim Chester disease is a very rare histiocytic disorder characterised by tissue infiltration by lipid laden histiocytes. The most common presentation is bone pains typically involving the long bones. Over time almost 50% of the patients develop extraosseous involvement. The prognosis depends on the extent and distribution of the extraskeletal manifestations. Cardiovascular involvement is seen in up to 40% of the patients and the most common manifestations are periaortic fibrosis and pericardial involvement. Respiratory distress, extensive pulmonary fibrosis, and cardiac failure are the most common causes of death in these patients. Cardiac tamponade has also been documented to cause death in these patients. We describe a patient of Erdheim Chester disease who presented with recurrent and very rapidly occurring cardiac tamponade in a short duration of time and benefited from timely recognition and management.	22572439

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Apr	Vnitr Lek - Czech	Interleukin-1 receptor blockade with anakinra provided cessation of fatigue, reduction in inflammation markers and regression of retroperitoneal fibrosis in a patient with Erdheim-Chester disease - case study and a review of literature	Adam Z, Szturz P, Bučková P, Cervinková I, Koukalová R, Rehák Z, Krejčí M, Pour L, Zahradová L, Hájek R, Král Z, Mayer J.	Interni hematoonkologicka klinika Lekarske fakulty MU a FN Brno. z.adam@fnbrno.cz	We describe a case of an Erdheim-Chester disease patient. First line chemotherapy treatment with 2-chlorodeoxyadenosine did not reduce fluorodeoxyglucose accumulation in pathological lesions. The patient had continuously increased CRP values of 17-20 mg/l. The disease continued to cause subfebrile temperatures and significant fatigue that made the patient to spend most of the daytime in bed. To manage the permanently increased inflammation markers, we decided to start treatment with anakinra, successfully used in some other autoinflammatory diseases (e.g. Schnitzler syndrome). We have now been able to evaluate the first 6 months of treatment. Daily subcutaneous administration of anakinra (KineretTM 100 mg daily) led to normalization of CRP values, cessation of subfebrile temperatures and, importantly, significant reduction of fatigue. Time periods the patient was able to spend out of the bed increased significantly. Consequent to the reduced fatigue, the patient was able to perform basic household tasks he was unable to undertake without treatment. After 3 months of treatment, fatigue of the same intensity returned following a short interruption of therapy. The CRP values went up again to 12 mg/l. CRP value returned back to norm and fatigue ceased after re-initiation of daily Kineret injections. Objective treatment response was assessed by measuring the degree of fluorodeoxyglucose accumulation in pathological bone lesions. PET-CT was performed before and 3 and 6 months after anakinra initiation. Intensity of accumulation did not change significantly after the first 3 months of therapy but decreased after 6 month therapy. Follow up CT of abdominal cavity was performed at the end of the 6th month of treatment. Presented CT images from before and 6 months after the treatment evidence an obvious reduction in fibroid changes in the retroperitoneum. Daily administration of anakinra to a patient with active Erdheim-Chester disease significantly reduced intensity of fatigue and improved quality of life, led to a reduction in inflammatory markers and regression in retroperitoneal fibrotization.	22559807
2012 May	Brain Pathol	A 38-year old woman with a dural based lesion	Husain S, Alkhalidi HM, Raddaoui E.	Dept. of Pathology King Khalid Univ. Hospital, Riyadh, Saudi Arabia	Isolated intracranial xanthogranulomas arising from the dura mater are extremely rare.We present a case of a symptomatic large right frontoparietal dura based intracranial xanthogranuloma in a 38-year-old female. Xanthogranulomas are benign non-Langerhans cell histiocytic lesions. They are frequently described in the skin of infants and children. Extracutaneous manifestations especially in the central nervous system are highly uncommon. Dural xanthogranulomas usually arise in association with familial hypercholesterolemia, with Erdheim Chester disease (ECD), and with Weber-Christian disease. Our case however, had no such associations. In this report, the authors describe the clinical, radiological and microscopic presentation of this case and the differential diagnoses of intracranial xanthogranuloma.	22497617
2012 May	Prog Urol.	Erdheim-Chester disease: Report of a case and literature review	Prunel P, Verhoest G, Besnard S, Rohou T, Rioux-Leclercq N, Bensalah K.	Service d'urologie, hôpital Pontchaillou, CHU de Rennes, 2, rue Henri-Le-Guilloux, 35033 Rennes cedex, France Email: paul.prunel@gmail.com	The Erdheim-Chester disease is a rare non-Langerhans hystiocytose acquired in adults. It results from a xanthogranulomatous infiltration, consists of histioccytes foamy and is characterized by heterogeneous systemic manifestations. The most frequent clinical manifestations of the disease are the bone with a long bone uptake on bone scintigraphy99Tc (Dion et al., 2006 [1]) and urological damage with an array of pseudo retroperitoneal fibrosis. We report the case of a 64-year-old man in whom was founded in the course of acute obstructive renal disease with Erdheim-Chester pseudofibrose retroperitoneal. Article in French.	22515929

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Mar	J Neurol Surg A Cent Eur Neurosurg	Erdheim-chester disease-a rare differential diagnosis of eosinophilic granuloma: a case report	Platz R, Romeike BF, Pandey DK, Kalff R, Reichart R.	University Clinic Jena, Neurosurgical Clinic, Jena, Germany.	No abstract available.	22467484
2012 Mar 30	QJM.	Progressive dysphagia caused by Erdheim-Chester disease	Vermeiren P, Van Laecke S, Cuvelier C, De Loose D, Vanholder R.	Renal Division, Department of Pathology and Department of Gastroenterology, Ghent University Hospital, Belgium.	. No abstract available.	22466415
2012 Mar	Br J Radiol	Erdheim-Chester disease associated with intramedullary spinal cord lesion.	Takeuchi T, Sato M, Sonomura T, Itakura T.	Dr Morio Sato, Department of Radiology, Wakayama Medical University, 811-1 Kimiidera, Wakayamashi, Wakayama 641-8510, Japan. E-mail: morisato@mail.wakayama-med.ac.jp	Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis. We present a case of a 56-year-old male with ECD. As time progressed, involvement of the orbital fossa, cranial convexity, spinal cord, brain stem, thyroid, lung, retroperitoneum, lower extremity bones and skin were found. Previously reported cases reveal the frequency of ECD with spinal cord involvement is rare. Although this was a presumed diagnosis based on other lesions, our case is the first in which both intramedullary and epidural masses are present.	22391503
2012 Feb 29	Rheumatology (Oxford)	Whole-body MRI in Erdheim-Chester disease.	Arnaud L, Bach G, Zeitoun D, Drier A, Cluzel P, Grenier PA, Amoura Z, Haroche J.	Department of Internal Medicine, Department of Radiology and Departement of Neuroradiology, AP-HP, Groupe Hospitalier Pitié-Salpêtrière, UPMC Univ Paris, Paris, France.	No abstract available.	22378719
2011 Dec 10	Nihon Naika Gakkai Zasshi	Case report; an autopsy case of Erdheim-Chester disease involving the lung and heart	Suzuki S, Matsuura T, Fukuda M, Take M, Kubo S, Yoshimoto T.	Department of Internal Medicine, Asoka Hospital, Japan.	Japanese. No abstract available	22338895

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Jan 31	Semin Arthritis Rheum	Treatment of Erdheim-Chester Disease with Long-Term High-Dose Interferon-α	Hervier B, Arnaud L, Charlotte F, Wechsler B, Piette JC, Amoura Z, Haroche J.	Department of Internal Medicine, APHP, French Reference Center for Auto-Immune Diseases, Hôpital Pitié-Salpêtrière, Paris, France; UPMC University of Paris 06, Paris, France	OBJECTIVES: Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis, characterized by a foamy CD68+, CD1a- histiocyte tissue infiltration. Efficacy of standard doses of interferon-α-2a (IFNα) has been suggested in a small series but with variation, depending on the organs involved. Our aim was to report our single-center experience about the use of high-dose IFNα in ECD. METHODS: Twenty-four ECD patients have received high-dose IFNα (IFNα ≥18 mIU/wk or pegylated-IFNα ≥180 μg/wk). IFNα efficacy was evaluated clinically and morphologically using a standardized protocol (median follow-up 19 months). RESULTS: Indication for treatment was central nervous system and/or heart involvement (n = 20), exophthalmos (n = 1), and standard-dose IFNα inefficacy (n = 3). High-dose IFNα was effective in 16 patients (67%) with improvement (n = 11, 46%) and stabilization (n = 5, 21%). Late and gradual improvement was observed during prolonged follow-up in most patients. The efficacy of high-dose IFNα was dependent on the organs involved: central nervous system and heart improvement or stabilization occurred in 7/11 (64%) and 11/14 (79%) patients, respectively. Six patients (25%) worsened. High doses of IFNα were well-tolerated: 13 (54.2%) patients had side effects but treatment interruption was infrequent (n = 3, 12.5%). CONCLUSIONS: High-dose IFNα may be effective in severe ECD. Improvement may be slow, and high-dose IFNα treatment should be prolonged	22300602
2012 Feb 1	Am J Respir Crit Care Med.	Erdheim chester disease: an unusual fluid overload mimic	Joshi M, Olman M.	-	-	22298365
2011 Dec 8	Int J Surg Case Rep.	Erdheim-Chester disease: The role of video-assisted thoracoscopic surgery in diagnosing and treating cardiac involvement.	Egan A, Sorajja D, Jaroszewski D, Mookadam F.	Cardiovascular Diseases Mayo Clinic Arizona 13400 E Shea Blvd Scottsdale, AZ 85259-5499 United States. Tel.: +1 480 301 6907 fax: +1 480 301 8018. Email: mookadam.farouk@mayo.edu	Erdheim-Chester disease is a rare, non-Langerhans histiocytosis in which pericardial involvement is diagnosed with increasing frequency and is associated with high mortality rates. PRESENTATION OF CASE: A 53-year-old woman presented with progressive exertional dyspnea and pericardial effusion was discovered. Further investigations revealed the presence of a diffuse, infiltrating process and a diagnosis of Erdheim-Chester disease was made. An emergent pericardiocentesis by subxiphoid approach was completed but recurrent drainage obviated removal of the pigtail catheter. A pleuro-pericardial window was placed using video-assisted thoracoscopic surgery (VATS) and analysis of the resected specimen confirmed pericardial involvement. DISCUSSION: In this case, high pericardial fluid output demanded definitive treatment of the pericardial effusion. Traditionally this would be completed via thoracotomy. VATS is a minimally invasive alternative which permits exploration of the thoracic cavity and the creation of a pleuropericardial window. CONCLUSION: We describe, for the first time, the successful use of VATS for both diagnostic confirmation and therapeutic relief of recurrent pericardial fluid drainage due to pericardial involvement by Erdheim-Chester disease.	22288060

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Dec 27	Cytojournal	Cytomorphology of Erdheim-Chester disease presenting as a retroperitoneal soft tissue lesion	Purgina B, Jaffe R, Monaco SE, Khalbuss WE, Beasley HS, Dunn JA, Pantanowitz L.	Ronald Jaffe, MB.BCh Department of Pathology Children's Hospital of Pittsburgh 4401 Penn Avenue Pittsburgh PA 15224 USA e-mail: Ronald.jaffe@chp.edu	Erdheim-Chester disease (ECD) is a rare, multisystem disorder of macrophages. Patients manifest with histiocytic infiltrates that lead to xanthogranulomatous lesions in multiple organ systems. The cytologic features of this disorder are not well characterized. As a result, the cytologic diagnosis of ECD can be very challenging. The aim of this report is to describe the cytomorphology of ECD in a patient presenting with a retroperitoneal soft tissue lesion. A 54-year-old woman with proptosis and diabetes insipidus was found on imaging studies to have multiple intracranial lesions, sclerosis of both femurs and a retroperitoneal soft tissue mass. Fine needle aspiration (FNA) and a concomitant core biopsy of this abnormal retroperitoneal soft tissue revealed foamy, epithelioid and multinucleated histiocytes associated with fibrosis. The histiocytes were immunoreactive for CD68, CD163, Factor XIIIa and fascin, and negative for S100, confirming the diagnosis of ECD. ECD requires a morphologic diagnosis that fits with the appropriate clinical context. This case describes the cytomorphologic features of ECD and highlights the role of cytology in helping reach a diagnosis of this rare disorder.	22279491
2011 Nov 10	Nihon Naika Gakkai Zasshi	Case report; a case of cardiac tumor diagnosed with Erdheim-Chester disease	Mochiduki N, Iida K, Kusuhara M, Kotani I.	Division of Cardiology, Shizuoka Cancer Center Hospital, Japan.	Japanese. No abstract available.	22250424
2012 Feb	Clin Nucl Med	F-18 FDG PET/CT Detects Muscle Involvement in Erdheim-Chester Disease	Ambrosini V, Savelli F, Merli E, Zompatori M, Nanni C, Allegri V, Fanti S.	Department of Nuclear Medicine, S. Orsola-Malpighi University Hospital, Bologna, Italy	A case of Erdheim-Chester disorder, a rare non-Langerhans' cell histiocytosis, was referred for restaging by F-18 FDG PET/CT more than 10 years after initial diagnosis. The patient presented diabetes insipidus, hypergondotropic hypogonadism, and osteosclerotic lesions. Previous bone scintigraphy documented pathognomonic long bones' involvement. Chronic steroid and hormone replacement therapy was administered, and the patient was asymptomatic. F-18 FDG PET/CT was useful for disease restaging at cardiac and soft tissues level.	22228352
2011 Aug;	Rev Med Chil	Erdheim-Chester disease: Report of one case	Vega J, Cisternas M, Bergoeing M, Espinosa R, Zapico A, Chadid P, Santamarina M.	Dr. Jorge Vega Stieb 5 Norte 1035, Viña del Mar, Chile. Fono: 56-32-2974237 Fax: 56-32-2970050 E-mail: jvegastieb@gmail. com	We report a 76-year-old male who was admitted due to progressive congestive heart failure lasting several months. An echocardiogram showed a large pericardial effusion with early signs of pericardial tamponade and an irregular surface suggestive of cancer infi ltration. The patient was operated, creating a pericardial window and draining 1,200 ml of a brownish yellow fl uid with abundant cellularity. Pericardial biopsy showed infi ltration by CD68 (+), CD1a (-) and S100 (-) cells. Twenty-eight months earlier, due to fatigue, dyspnea, and a non-specifi c inflammatory process, an enhanced-contrast-scan showed that aorta was coated with a hypodense tissue that began near the aortic valve and extended until the inferior mesenteric artery, with stenosis of the left subclavian, celiac axis, renal and upper mesenteric arteries. An angioplasty and stent placing was carried out in the last two arteries. Both kidneys had the appearance of “hairy kidneys”. A bone scan showed increased uptake in femurs and tibiae and X-ray examination showed osteosclerosis in metaphysis and diaphysis. The diagnosis of Erdheim-Chester disease (non-Langerhans-cell histiocytosis) was made and the patient was treated with steroids and methotrexate. (article in Spanish)	22215336

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2012 Jan;24	Curr Opin Rheumatol.	Langerhans cell histiocytosis and Erdheim-Chester disease.	Wilejto M, Abla O.	Division of Haematology/Oncology, Department of Paediatrics, The Hospital for Sick Children, Toronto, Ontario, Canada.	PURPOSE OF REVIEW: To provide an updated overview of the pathogenesis and treatment of Langerhans cell histiocytosis (LCH) and Erdheim-Chester disease (ECD). RECENT FINDINGS: There is ongoing debate as to the exact pathogenesis of these disorders and their classification as reactive versus neoplastic. Proinflammatory cytokines are known to play a role in both LCH and ECD and strengthen the hypothesis that, at least in part, they are disorders of immune dysregulation. The recent discovery of activating mutations in the proto-oncogene BRAF in a subset of LCH patients suggests that LCH is in fact a neoplastic disorder. Understanding of the mechanisms that promote proliferation and migration of histiocytes has led researchers to explore targeted immune-modulatory therapies for ECD. Similarly for LCH, alternative chemotherapeutic agents and reduced-intensity hematopoietic stem cell transplant are being evaluated for refractory disease. SUMMARY: More research is needed to better understand the cause of these disorders and may help in identifying new targeted therapies, particularly for patients with refractory or relapsed disease. Multinational trials are ongoing for LCH and are urgently needed for ECD.	22157416
2011 Dec	J Am Coll Cardiol	Interferon-alpha in cardiac erdheim-chester disease.	Haroche J, Sciarra A, Balzarini L, Fiamengo B, Amoura Z, Graziani G.	Monti L, I.R.C.C.S. Istituto Clinico Humanitas, Rozzano, Italy; University of Milan, School of Medicine, Milan, Italy.		22152958
2011 Nov 13	Curr Opin Rheumatol	Erdheim-Chester disease	Haroche J, Arnaud L, Amoura Z.	Department of Internal Medicine and French Reference Center for Rare Autoimmune and Systemic Diseases, Assistance Publique-Hôpitaux de Paris, Pitié-Salpêtrière Hospital	PURPOSE OF REVIEW: Erdheim-Chester disease (ECD) is a rare, non-Langerhans form of histiocytosis first described in 1930 with a wide range of manifestations. The number of new cases has dramatically increased over the past 10 years because of the better recognition of this condition. The natural evolution is variable, but the spontaneous prognosis is severe. In this review, we describe the relevant clinical, radiological, prognostic, and therapeutic features of this orphan disease. RECENT FINDINGS: Compelling evidence demonstrates the efficacy of treatment by interferon alpha (IFNα) which has been reported to be a major independent predictor of survival among ECD patients. Alternative treatments remain to be defined. Recent studies have highlighted the central nervous system involvement as an independent predictor of death. Pathophysiology is better understood with a complex network of cytokines and chemokines and a systemic immune Th-1-oriented perturbation. SUMMARY: ECD, although a rare and orphan disease, has been overlooked and numerous new cases are currently diagnosed because of general better knowledge of this histiocytosis. First-line treatment is IFNα. We have recently described a unique cytokine signature that may provide further clues to understand the pathogenesis of ECD, as well as provide new tools for diagnosis and targeted therapy.	22089098

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Sep;14	Hell J Nucl Med	(18)F-FDG positron emission tomography/computed tomography and (99m)Tc-MDP skeletal scintigraphy in a case of Erdheim-Chester disease	Asabella AN, Cimmino A, Altini C, Notaristefano A, Rubini G.	Nuclear Medicine Unit-Di.M.I.M.P., University of Bari, Piazza G. Cesare, 11, 70124 Bari, Italy. a.niccoli@mednucl.uniba.it	Erdheim-Chester disease (ECD) is a rare form of non-Langerhan's cell histiocytosis with unknown aetiology, is charaterized by systemic xanthogranulomatous infiltrative disease. The typical ECD diagnostic triad is bone pain, diabetes insipidus and bilateral exophthalmos. A 24 years old man came at our attention for polydipsia with nocturnal and diurnal polyuria, anorexia, febrile episodes, and arthromyalgia especially in the knees. Physical examination showed bilateral periorbital xanthelasma. Blood exams showed increase of plasma osmolarity, haematocrit, sodium and urea and decrease of potassium. Urine exams showed just decreased urine specific gravity, suggestive for central diabetes insipidus (CDI). Brain magnetic resonance with gadolinium enhancement showed the presence of multiple hyperintense lesions expecially in neurohypophysis (swollen and with markedly contrast enhancement). Two weeks later, whole-body plus lower limbs 18-fluorine-labelled 2-deoxy-2-fluoro-D-glucose positron emission tomography/computed tomography ((18)F-FDG PET/CT) was performed. Uptake of (18)F-FDG was observed in the upper portion of the midbrain area (SUV(max) 7.1) and the pituitary gland (SUV(max) 7.3), and diffuse bone marrow uptake of (18)F-FDG in the proximal epiphysis and metaphysis of both humeri and thigh bones (SUV(max) 6.5), shoulder blades, pelvis bones and the L2 vertebral body (SUV(max) 3.9). This (18)F-FDG PET/CT confirmed the presence of brain lesion seen in MRI , the absence of visceral lesions, but also showed the presence of an atypical bone uptake of (18)F-FDG, leading to the suspision of ECD. A technetium-99m-methyl-diphosphonate skeletal scintigraphy ((99m)Tc-MDP) scan showed diffuse uptake of the radiopharmaceutical, in the diaphysis of long bones and in the left portion of the body and the spinous process of L2. Considering the difficulties of an osteomedullary or brain biopsy, biopsy was performed on a right anterior thoracic cutaneous xanthelasma. Histology showed lipid-laden histiocytes (CD1a-, CD68+, S-100 protein -) with small nuclei, Touton giant, lymphocytic infiltrates, eosinophils and fibrosis, ECD gold standard patterns as reported in literature. The patient was discharged with the diagnosis of ECD with central nervous system (CNS) manifestations, and treatment started. The diagnosis can be lead by the most charateristic bone findings of symmetrical osteosclerosis of the long bones, especially the lower limbs (tibia and fibula), involving metaphyses and diaphyses but sparing epiphyses. About half of all ECD patients may experience extraskeletal manifestations, including CNS. Visceral involvement in ECD is not specific, and this enforces the diagnostic value of skeletal imaging findings. Furthermore xanthomas can be found at any location on the skin, especially the eyelids as in our patient. For visceral involvement, CT is most useful, while MRI is more sensitive for CNS lesions. Involvement of CNS may be frequently revealed clinically by diabetes insipidus. Few case reports have shown that (18)F-FDG PET/CT scanning could be useful in assessing the extension of ECD lesions. Both radiography and (99m)Tc-MDP skeletal scintigraphy may reveal osteosclerosis of the long bones, which is a typical finding in ECD. The typical bone pattern of (18)F-FDG PET/CT scan is specific for ECD and (99m)Tc-MDP skeletal scintigraphy may be performed in patients in whom initial (18)F-FDG PET/CT scans present the possibility of ECD diagnosis. Others reported that (18)F-FDG PET/CT scans had good sensitivity (66.7%) and specificity (92.3%) as compared with MRI of the CNS involvement or lesions. In conclusion, the (18)F-FDG PET/CT scan and the (99m)Tc-MDP scan depicted many of the most relevant lesions of ECD for the initial assessment of ECD in our patient.	22087457

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Nov	Radiographics	Sclerosing bone dysplasias: review and differentiation from other causes of osteosclerosis	Ihde LL, Forrester DM, Gottsegen CJ, Masih S, Patel DB, Vachon LA, White EA, Matcuk GR Jr.	Department of Radiology, Keck School of Medicine, University of Southern California, 1500 San Pablo St, 2nd Floor Imaging, Los Angeles, CA 90033-5313	Sclerosing bone dysplasias are skeletal abnormalities of varying severity with a wide range of radiologic, clinical, and genetic features. Hereditary sclerosing bone dysplasias result from some disturbance in the pathways involved in osteoblast or osteoclast regulation, leading to abnormal accumulation of bone. Several genes have been discovered that, when disrupted, result in specific types of hereditary sclerosing bone dysplasia (osteopetrosis, pyknodysostosis, osteopoikilosis, osteopathia striata, progressive diaphyseal dysplasia, hereditary multiple diaphyseal sclerosis, hyperostosis corticalis generalisata), many of which exhibit similar pathologic mechanisms involving endochondral or intramembranous ossification and some of which share similar underlying genetic defects. Nonhereditary dysplasias include intramedullary osteosclerosis, melorheostosis, and overlap syndromes, whereas acquired syndromes with increased bone density, which may simulate sclerosing bone dysplasias, include osteoblastic metastases, Paget disease of bone, Erdheim-Chester disease, myelofibrosis, and sickle cell disease. Knowledge of the radiologic appearances, distribution, and associated clinical findings of hereditary and nonhereditary sclerosing bone dysplasias and acquired syndromes with increased bone density is crucial for accurate diagnosis.	22084176
2011 Nov 11	Eur J Echocardiogr	Cardiac involvement in Erdheim-Chester disease: echocardiographic appearance and value of cardiac MRI	Merli E, Savelli F, Lovato L, Zompatori M.	Department of Cardiology, Ospedale per gli Infermi, viale stradone 9, 48018, Faenza, Ravenna, Italy. elisamerli@libero.it		22080452

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Nov	Ann Dermatol Venereol	Associated Langerhans cell histiocytosis and Erdheim-Chester disease	Marchal A, Cuny JF, Montagne K, Haroche J, Barbaud A, Schmutz JL.	Service de dermatologie, hôpitaux de Brabois, CHU Nancy, bâtiment des spécialités médicales Philippe-Canton, rue du Morvan, 54500 Vandœuvre-lès-Nancy, France.	BACKGROUND: Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis with multiple organ involvement affecting middle-aged adults. A case of ECD associated with Langerhans cell histiocytosis (LCH) is reported herein. CASE REPORT: A 75-year-old woman presented maculopapular skin lesions on her trunk, associated with constrictive pericarditis and pleurisy present for 1year. The skin biopsy militated in favour of LCH since it revealed a histiocytic infiltrate with a positive CD1a marker at immunohistochemistry (IHC). The association with ECD was diagnosed on the basis of pericarditis, periaortitis, pleurisy, pulmonary involvement and retroperitoneal fibrosis. The patient was treated with interferon-α2a with good initial results, but died from septic shock a year and a half later, a few months after discontinuing interferon due to poor tolerability. DISCUSSION: The clinical, radiographic and histological arguments in favour of ECD clearly differ from those for LCH. However, as already reported, the two illnesses may be associated, thus underlining the possible existence of a link between these two histiocytic proliferations emanating from the same medullary precursor. Two hypotheses have been advanced in an attempt to explain this association: the first involves a stimulus that might lead to independent proliferation of the two cell lines while the second suggests the existence of a transformation pathway from one form of proliferation to the other. CONCLUSION: Screening for associated ECD should be routinely performed in patients presenting LCH with signs evocative of ECD.	22078035

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011	Klin Onkol	Partial regression of CNS lesions of Erdheim-Chester disease after treatment with 2-chlorodeoxadenosine and their full remission following treatment with lenalidomide].	Adam Z, Sprláková A, Rehák Z, Koukalová R, Szturz P, Krejcí M, Pour L, Zahradová L, Cervinek L, Kren L, Moulis M, Hermanová M, Mechl M, Prásek J, Hájek R, Král Z, Mayer J.	Interní hematoonkologická klinika, LF MU a FN Brno. z.adam@fnbrno.cz	INTRODUCTION: Erdheim-Chester disease is a very rare syndrome affecting adult population. It typically causes hyperostosis of long bones, retroperitoneal fibrosis and widening of the aortic wall. Patients frequently suffer from disease-associated fevers and pain in the lower limbs. No guidelines are available for the treatment of this rare ailment. Therefore, we describe our experience with lenalidomide in a patient with poor treatment response to 2-chlorodeoxyadenosine. CASE: Diabetes insipidus and neurological problems developing over 4 years were the first signs of the disease. The disease was diagnosed from histology of the bone marrow extracted from the ilium. At diagnosis, the patient had multiple infiltrates in the brain, widened wall of the thoracic and abdominal aorta, fibrotic changes to retroperitoneum and typical hyperostosis of the long bones of lower limbs with high accumulation of technetium pyrophosphate as well as fluorodeoxyglucose. First line treatment involved 2-chlorodeoxyadenosine 5 mg/m2 s.c. for 5 consecutive days every 28 days. There was no clear treatment response identifiable on the MR scan of the brain following the third cycle and thus 4th-6th cycle consisted of 2-chlorodexyadenosine 5 mg/m2 + cyclophosphamide 150 mg/m2 + dexamethasone 24 mg day 1-5 every 28 days. After the 6th cycle, MR showed partial regression of the brain lesions. PET-CT showed an increased accumulation of fluorodeoxyglucose in bone lesions. Second line treatment involved lenalidomide 25 mg/day days 1-21 every 28 days. Lenalidomide tolerance was excellent; the number of neutrophils and thrombocytes was within the physiological range throughout the treatment period. Follow-up MR showed complete remission of the brain lesions, while follow-up PET-CT showed further increase in fluorodeoxyglucose accumulation in the bones of lower limbs. CONCLUSION: Treatment with 2-chlorodeoxyadenosine-based regimen provided partial remission of Erdheim-Chester disease lesions in the brain, while treatment with lenalidomide resulted in complete remission of these lesions. Fluorodeoxyglucose continues to accumulate in the long bones of lower limbs. We are unable to elucidate the reasons for complete remission of the disease in the brain as per the MR and its progression in the long bones according to PET-CT. Further testing of lenalidomide in the treatment of this disease is required to support further use of this perspective treatment option.	22070019

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Nov	J Clin Oncol.	Role of iconographic examinations in the treatment algorithm in erdheim-chester disease.	Aouba A, Bienvenu B, Launay D, Hermine O.	Department of Adult Haematology Hôpital Necker Enfants-Malades, Université de Paris Descartes, Assistance Publique-Hôpitaux de Paris, 149 rue de Sèvres, 75015 Paris, France; achaouba@sfr.fr or achille.aouba@nck.aphp.fr.	We read with interest the article by Balink et al1 on a patient with Erdheim-Chester disease (ECD) who had typical skeletal features from technetium-99 bone scintigraphy and a positive diagnosis obtained by tibial biopsy. Using positron emission tomography (PET)/computed tomography (CT) with fluorodeoxyglucose ([18F]FDG), the authors found corresponding bone lesions and abnormal bifocal uptake in the lungs, suggestive of infiltrative change. On the basis of this extraskeletal involvement, the authors chose imatinib mesylate therapy. However, we would like to draw attention to the role of iconographic exams for the diagnosis and choice of treatment in ECD. When listing the different ECD treatments, the authors omitted anakinra, which has recently demonstrated good outcomes in two patients, including a complete response.4 Anakinra, a recombinant form of interleukin (IL) -1RA, specifically targets the proinflammatory interleukine-1, which is probably overexpressed in ECD.4 Indeed, anakinra shares specific mechanisms of action with interferon (INF) therapy, as INF increases monocytic production of endogenous IL-1RA. INF is the drug most used for ECD and improves survival.5 Because anakinra exhibits a better tolerance profile than INF therapy, anakinra seems to be a new and promising treatment for ECD. By successively blocking IL-1, IL-6, and systemic inflammation,4,6 both anakinra and INF therapy could have corrected the severe anemia of chronic disease observed in this patient. Considering the risk of severe cytopenia and bleeding with imatinib and INF therapy, anakinra should have been the preferential drug in this patient, who also suffered intermittent rectal bleeding. Finally, whatever the result of the PET-[18F]FDG scan, this patient would have needed active systemic treatment because of severe systemic repercussions of the chronic inflammatory syndrome. Only one case report has described a successful outcome with imatinib for ECD.3 The use of this drug is limited, and results are disappointing for this disease; other data are available on only six other patients with ECD selected for tyrosine kinase overexpression who received imatinib: two had a stable outcome and four had progressive disease.7 Therefore, INF therapy is largely used in first-line therapy for ECD, rather than imatinib.5,7,8 Moreover, the argument of Balink et al to preferentially use imatinib because of extraskeletal involvement is surprising, as most ECD patients exhibit extraskeletal involvement. For patients who exhibit isolated, stable bone localization without any systemic manifestation, a single nonsteroidal anti-inflammatory drug therapy should be sufficient.4 Taken together, even though imatinib may have some efficacy for a subset of ECD patients, we are surprised at these authors' recommendation for its use as first-line therapy for ECD. In addition, this recommendation does not appear in the article by Janku et al3 to which Balink et al refer. Corticosteroids symptomatic efficacy is often only partial and transient. The use of bisphosphonate is also anecdotal and is probably only symptomatic. In contrast, anakinra has demonstrated dramatic and complete control of all constitutional symptoms.4 To summarize, the diagnostic use of bone scintigraphy, as referred to in the title of the Balink et al article, and the staging help of PET-[18F]FDG scan, are already well known. In contrast, the activation status of receptor tyrosine kinase and the outcome of imatinib mesylate therapy assessed notably with PET-[18F]FDG should be interesting additive data to report on this rare disease.	22025165

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Sep 29	AJNR Am J Neuroradiol	Erdheim-Chester Disease of the Central Nervous System: New Manifestations of a Rare Disease	Sedrak P, Ketonen L, Hou P, Guha-Thakurta N, Williams MD, Kurzrock R, Debnam JM.	J. Mathew Debnam, MD, University of Texas MD Anderson Cancer Center, 1400 Pressler Blvd, Unit 1482, Houston, Texas 77030; e-mail: Matthew.Debnam@mdanderson.org	**BACKGROUND AND PURPOSE:** ECD is a rare non-Langerhans-cell histiocytosis, which can involve the CNS; therefore, CNS imaging findings have been described in only a small number of patients. To gain additional insight into the CNS manifestations of ECD, we reviewed the findings on imaging of the brain, head and neck, and spine in patients with ECD who presented to our institution. Here, we illustrate manifestations that have not, to our knowledge, been previously described. **MATERIALS AND METHODS:** CT, MR imaging, and PET/CT studies of the brain, maxillofacial region, and spine were reviewed in 11 patients with ECD. **RESULTS:** Four new manifestations of ECD were present, including the following: a stellate appearance of intracranial extra-axial lesions, ependymal enhancement along the lateral ventricle with deep linear extension to the lentiform nucleus, irregular enhancement in the pons, and diffuse involvement of the vertebral column on PET/CT. **CONCLUSIONS:** ECD has a variety of imaging appearances in the CNS, including new manifestations described herein. Neuroradiologists should be aware of these manifestations to avoid mistaking them for other disease processes.	21960492
2011 Sep	Dtsch Med Wochenschr	Recurrent pericardial effusion as first manifestation of Erdheim-Chester disease	Lutz SZ, Schmalzing M, Vogel-Claussen J, Adam P, May AE.	Abteilung für Endokrinologie, Diabetes, Nephrologie, Angiologie und Klinische Chemie, Medizinische Universitätsklinik, Eberhard-Karls-Universität, Tübingen Germany	**History and admission findings:** A 65-year-old woman presented with reduced general condition and dyspnoea that was progressive over the last months. Clinical findings revealed an exophthalmus on the right, xanthelasm and mild peripheral oedema. Previously, a pericardiocentesis had been performed due to a large pericardial effusion. A previous CT scan showed a mass attached to the pericardium extending through the atrio-ventricular groove and a thickened aorta. In addition, a retroperitoneal fibrosis and an occlusion of both Aa. iliacae internae were found. **Investigations:** The ECG showed sinus rhythm. Laboratory findings demonstrated a microcytic anemia and a renal failure. Chest radiography showed a large cardiac silhouette, while the transthoracic echocardiography revealed a recurrent large pericardial effusion. A PET/CT scan of the chest and abdomen showed a tissue infiltration of the retroperitoneal structures, a mass surrounding the right coronary artery and the right orbita. Finally, a femur biopsy confirmed the diagnosis of Erdheim-Chester disease. **Diagnosis,** treatment and course: With the diagnosis Erdheim-Chester disease we started a high dose immunsuppressive therapy using glucocorticoids and interferon-a. Tumour size slightly decreased during the following 2 months, however the patient developed a severe urosepsis and died from multiorgan failure. **Conclusions:** We report a case of an Erdheim-Chester disease with cardiovascular involvement primarily diagnosed due to a recurrent large pericardial effusion. In case of cardial tumors with interatrial septum or coronary artery involvement together with cerebral manifestations, an Erdheim-Chester disease should be taken into account.	21935854

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Sep 21	Respiration	Erdheim-Chester Disease Presenting with Pneumothorax	Yamaguchi M, Shiota T, Kobashi Y.	Masafumi Yamaguchi, MD, PhD Department of Cardiovascular and Respiratory Medicine, Shiga University of Medical Science, Seta Tsukinowa-cho Otsu, Shiga 520-2192 (Japan) Tel. +81 77 548 2213, E-Mail myama3265@gmail.com	Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis most commonly characterized by symmetrical skeletal involvement and may present with pulmonary involvement leading to chronically progressive pulmonary symptoms. Characteristics on chest radiography include non-specific findings of diffuse interstitial and pleural thickening, micronodules, ground-glass opacities and parenchymal condensation as a result of infiltration by lipid-laden histiocytes. We present the case of a 50-year-old man with ECD presenting with acute pulmonary symptoms due to rupture of a large cystic lesion with resultant pneumothorax. He was brought by ambulance to our hospital, complaining of acute anterior chest pain and severe dyspnea. Chest radiography showed right-sided pneumothorax with a collapsed lung, a large, left-sided cystic lesion in the upper lung field and accentuated interstitial markings. Bullectomy and surgical biopsy were performed, demonstrating histologically histiocytic infiltrates that were strongly positive for CD68, but negative for S-100 protein and CD1a. Subsequent systemic examinations indicated widespread symmetrical skeletal involvement, leading to a definitive diagnosis of ECD.	21934273
2011 Aug 26	Arthritis Rheum.	Treatment of pediatric erdheim-chester disease with interleukin-1 targeting drugs	Tran T, Pariente D, Lecron J, Delwail A, Taoufik Y, Meinzer U.	Department of Pediatrics, Pediatric Rheumatology, CEREMAI, Hôpital Bicêtre Université Paris Sud, Le Kremlin Bicêtre, France; INSERM U-1012 Université Paris-Sud, Bicêtre, France. tu-anh.tran@bct.aphp.fr.	No abstract available.	21898344
2011 Aug 31.	Pediatr Radiol.	Erdheim-Chester disease with multisystem involvement in a 4-year-old	Song SY, Lee SW, Ryu KH, Sung SH.	Department of Radiology, Ewha Womans University School of Medicine, Seoul, South Korea, littlessook@hanmail.net	Erdheim-Chester disease is a rare form of non-Langerhans histiocytosis of unknown origin occurring mainly in adults. It is extremely rare in children. We report a case of a 4-year-old boy with Erdheim-Chester disease that initially presented as hemifacial palsy and bone pain with multisystem involvement. We describe radiographic findings of bones that show characteristic bilateral symmetrical osteosclerosis with atypical osteolytic lesions in addition to CT findings for pulmonary involvement and MR findings for intracranial lesions.	21879308

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Aug 9	Annales de Dermatologie et de Vénéréologie	Associated Langerhans cell histiocytosis and Erdheim-Chester disease	A. Marchala, J.-F. Cunya, K. Montagneb, J. Harochec, A. Barbauda, J.-L. Schmutz	Service de dermatologie, hôpitaux de Brabois, CHU Nancy, bâtiment des spécialités médicales Philippe-Canton, rue du Morvan, 54500 Vandœuvre-lès-Nancy, France	Background Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis with multiple organ involvement affecting middle-aged adults. A case of ECD associated with Langerhans cell histiocytosis (LCH) is reported herein. Case report A 75-year-old woman presented maculopapular skin lesions on her trunk, associated with constrictive pericarditis and pleurisy present for 1 year. The skin biopsy militated in favour of LCH since it revealed a histiocytic infiltrate with a positive CD1a marker at immunohistochemistry (IHC). The association with ECD was diagnosed on the basis of pericarditis, periaortitis, pleurisy, pulmonary involvement and retroperitoneal fibrosis. The patient was treated with interferon-α2a with good initial results, but died from septic shock a year and a half later, a few months after discontinuing interferon due to poor tolerability. Discussion The clinical, radiographic and histological arguments in favour of ECD clearly differ from those for LCH. However, as already reported, the two illnesses may be associated, thus underlining the possible existence of a link between these two histiocytic proliferations emanating from the same medullary precursor. Two hypotheses have been advanced in an attempt to explain this association: the first involves a stimulus that might lead to independent proliferation of the two cell lines while the second suggests the existence of a transformation pathway from one form of proliferation to the other. Conclusion Screening for associated ECD should be routinely performed in patients presenting LCH with signs evocative of ECD.	-

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Jun	Vnitr Lek	Successful treatment of Erdheim-Chester disease by 2-chlorodeoxyadenosine-based chemotherapy. Two case studies and a literature review	Adam Z, Koukalová R, Sprláková A, Rehák Z, Cervinek L, Szturz P, Krejcí M, Pour L, Zahradová L, Moulis M, Prásek J, Chaloupka R, Hájek R, Mayer J.	SourceInterní hematoonkologická klinika Lékarské fakulty MU a FN Brno. z.adam@fnbrno.cz	[Article in Czech] Abstract INTRODUCTION: Erdheim-Chester disease is an extremely rarely occuring condition and thus an optimal treatment is not known. Two new cases have been diagnosed in our centre in 2008 and 2009. Both patients had diabetes insipidus, B symptoms (subfebrile to febrile states) and pain in long bones of lower limbs. CASE STUDIES: Imaging showed high accumulation of fluorodeoxyglucose as well as Tc-pyrophosphate in long bones of lower as well as upper limbs, aortic wall thickening with periaortic fibrosis and perirenal fibrosis. In addition, one of the patients had multiple lesions in the brain. 2-chlorodeoxyadenosine 5 mg/m2 s.c. and cyclophosphamide 150 mg/m2 administered on days 1 to 5 in 28-day cycles were selected for the treatment of both patients. Dexamethasone 24 mg/day for 5 days was added to this treatment in the second patient. Six cycles of the treatment were planned. Both patients were prescribed bisphosphonates--zoledronate and clodronate, respectively. Treatment effect was assessed with PET-CT and MR. Following treatment completion, brain infiltrates were reduced to a small residuum in the first patient who did not anymore complain of leg pain. However, there was no reduction in fluorodeoxyglucose accumulation in bone lesions and thus treatment response was assessed as partial remission. This patient is currently receiving a second line treatment and treatment follow-up is 26 months from the diagnosis. Repeated PET-CTs in the second patient showed a significant reduction in accumulation of fluorodeoxyglucose in all pathological lesions. Febrile states and pain in long bones as well as pathological fatigue ceased after the treatment. Increased CPR and fibrinogen gradually returned to their normal levels. This response is assessed as complete remission. This patient's follow-up is 16 months from the diagnosis. CONCLUSION: Administration of 2-chlorodeoxyadenosine (5 mg/m2 s.c.) + cyclophosphamide (150 mg/m2 intravenously) and dexamethasone (24 mg/day) led to partial remission in one patient; nearly complete remission of CNS infiltrates but persistent elevation of fluorodeoxyglucose accumulation in bone lesions. Complete remission with a significant reduction in accumulation of fluorodeoxyglucose in all disease lesions with normalization of originally increased inflammatory markers and disappearance of all symptoms of the disease was achieved in the second patient.	21751544
2011 Jul-Aug	J Radiol.	Imaging features of osseous and extra-osseous involvement in Erdheim-Chester disease	Adib O, Baroth E, Perard L, Scoazec JY, Vervueren L, Aubé C, Willoteaux S.	SourceDépartement de radiologie, centre hospitalier universitaire d'Angers, 4, rue Larrey, 49933 Angers cedex 09, France	[Article in French] Erdheim-Chester disease is a rare form of systemic non-Langerhans cell histiocytosis characterized by infiltration by lipid-laden or foamy histiocytes. Osseous involvement, major diagnostic criteria, is constant and characteristic. It presents as metaphyseal and diaphyseal osteosclerosis, mainly affecting the long bones of the lower limbs. A few cases with axial skeleton involvement have been reported. Extra-osseous lesions may affect the retroperitoneum, lungs, skin, heart, brain and orbits. Prognosis depends mainly on the extra-osseous disease, mainly heart and lung involvement. Diagnosis is based on the combination of radiographic features, nuclear medicine features and nearly pathognomonic immunohistochemical profile.	21819909

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Jul 23	Rheumatol Int	Bilateral orbital Erdheim-Chester disease treated with 12 weekly administrations of VNCOP-B chemotherapy: a case report and a review of literature	Broccoli A, Stefoni V, Faccioli L, Agostinelli C, Spinardi L, Pastore Trossello M, Zinzani PL.	SourceInstitute of Hematology and Medical Oncology "L. e A. Seràgnoli", Policlinico "Sant'Orsola-Malpighi", University of Bologna, Via Massarenti, 9, 40138, Bologna, Italy, alessandro.broccoli@studio.unibo.it	Erdheim-Chester disease (ECD) is a non-Langerhans' cells histiocytosis of unknown etiology, which generally presents with long bones involvement, even if extraskeletal lesions may be frequently recognized. As a consequence of its rarity, there is no consensus concerning the best standard of care for affected patients. We present the case of a 53-year-old woman with bilateral orbital histologically documented ECD, presenting with an important thickening and swelling of the periorbital tissue and massive involvement of lateral rectal muscles, as documented by magnetic resonance. The patient was successfully addressed to 12 cycles of a weekly lymphoma-designed chemotherapy regimen, including etoposide, mitoxantrone, cyclophosphamide, vincristine, bleomycin, and prednisone (VNCOP-B regimen). Periorbital lesions reduced during the courses of chemotherapy, along with a regression to normal appearance of the extrinsic ocular musculature. This appears as an effective and well-tolerated first-line treatment option for ECD patients, due to the possibility of maintaining an adequate dose intensity, with also a concomitant continuous steroid administration.	21785962
2011 Aug	Eur J Neurol	Erdheim Chester disease presenting as slowly progressive cerebellar syndrome and asymptomatic widespread skeletal involvement	Tufan F, Myftiu B, Aygun D, Keles N, Haroche J, Hanagasi H, Gurvit H, Emre M, Besisik S.	SourceDivision of Geriatrics, Department of Internal Medicine, Istanbul School of Medicine, Istanbul University, Istanbu	Letter to the Editor	21749566
2011 Jun 16	MMW Fortschr Med	Which organs are involved here? Erdheim-Chester disease	Pfeil A, Jung C, Boettcher J, Wolf G, Hansch A.	SourceKlinik für Innere Medizin III, Universitätsklinikum Jena	Article in German	21717702
2011 Aug	Clin Nucl Med	Erdheim-chester disease: imaging-guided therapeutic approach	Della Torre E, Dagna L, Mapelli P, Mellone R, Grazia Sabbadini M.	San Raffaele Scientific Institute Università Vita-Salute San Raffaele School of Medicine Via Olgettina, 60 20132 Milano, ITALY Telephone: +39 02 2643 3872 Fax: +39 02 2643 3787 e-mail: dagna.lorenzo@hsr.it	Erdheim-Chester disease (ECD) is a rare form of systemic non-Langerhans cell histiocytosis with characteristic bone involvement. However, extraskeletal involvement occurs in approximately half of the patients. Because of its protean findings, the diagnosis of ECD is often delayed; thus, a clinical suspicion may prompt specific imaging studies to recognize suggestive signs of organ involvement. In this study, a case of a patient with ECD with representative progressive multisystemic involvement has been reported; although the final diagnosis was confirmed by histologic analysis, imaging studies with almost pathognomonic findings guided the diagnostic process and prompted different therapeutic approaches according to the localization of the disease.	21716027

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Jun 22	BMC Gastroenterol	Unusual Manifestation of Erdheim-Chester Disease	Pan A, Doyle T, Schlup M, Lubcke R, Schultz M	Gastroenterology Unit, Southern District Health Board, Dunedin, New Zealand Email: antony.pan@yahoo.com	BACKGROUND: Erdheim-Chester disease (ECD) is a rare multisystem non-Langerhans cell histiocytosis that is characterized histologically by xanthogranulomatous infiltrates and radiologically by symmetrical sclerosis of long bones. The xanthomatous process is characterized by prominent foamy histiocytes staining positive for CD68, occasionally for PS100 and negative for S100 and CD1a. Gastroenterological involvement is exceedingly rare. Case Presentation: This case report describes the case of a 69-year-old man who presented otherwise well to the gastroenterology department with unspecific abdominal symptoms, nausea, vomiting and weight loss. ECD involving the gastrointestinal tract was confirmed clinically, radiologically and histologically. CONCLUSION: Gastroenterological manifestation of ECD is rare but should be considered in the differential diagnosis in patients presenting with evidence of multi-organ disease and typical radiological features of Erdheim-Chester disease elsewhere.	21693070
2011 Jun 15	J Thorac Imaging	Erdheim-Chester Disease With Interatrial Septum Involvement	Raptis DA, Raptis CA, Jokerst C, Bhalla S	SourceMallinckrodt Institute of Radiology, Washington University, St. Louis, MO	Erdheim-Chester disease is an uncommon non-Langerhans cell histiocytosis with systemic manifestations. Most cases discuss radiologic findings once a pathologic diagnosis has already been established. We describe a patient with symptoms and no previously known diagnosis who was imaged with computed tomography, magnetic resonance imaging, and positron emission tomography. This case is unusual in that radiologic imaging demonstrated interatrial septum and diffuse cardiac involvement, in addition to the other characteristic lesions of Erdheim-Chester disease. The importance of this case to the radiologist is the expansion of the differential diagnosis of processes involving the interatrial septum and retroperitoneum.	21681117
2011 May	Case Rep Dermatol	Peculiar distribution of tumorous xanthomas in an adult case of erdheim-chester disease complicated by atopic dermatitis	Murakami Y, Wataya-Kaneda M, Terao M, Azukizawa H, Murota H, Nakata Y, Katayama I.	SourceDepartment of Dermatology, Osaka University Graduate School of Medicine, Osaka, Japan	Erdheim-Chester disease is a rare non-Langerhans form of histiocytosis with multiple organ involvement. Approximately 20% of patients have xanthoma-like lesions, usually on the eyelids. We report a case of Erdheim-Chester disease in a 32-year-old male who showed peculiar xanthomatous skin lesions and also had atopic dermatitis. His skin manifestations included ring-like yellowish tumors on his periorbital regions, rope necklace-like tumors on his neck, and spindle-shaped tumors on his right preauricular region and cubital fossas. He also had exophthalmos and diabetes insipidus. Chronic eczematous lesions were present on the flexor aspect of his extremities, and his serum eosinophil numbers and immunoglobulin E levels were elevated. A histological examination of his right neck tumor showed foamy macrophages and touton-type giant cells, which were positive for CD68 and CD163 and negative for S-100 and CD1a. We suggest that the complication of atopic dermatitis may have contributed to the uncommon clinical features in this case.	21677888
2011 Jun 13	Int J Cardiol	Cardiovascular involvement in Erdheim-Chester disease	Masci PG, Zampa V, Barison A, Lombardi M.	Cardiovascular Magnetic Resonance Imaging and §Cardiovascular Medicine Departments, Fondazione CNR/Regione Toscana 'G.Monasterio', Pisa, Italy		21676475

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Jul-Aug	Dimens Crit Care Nurs	Erdheim-chester disease: a case study and literature review	Andrysek K.		Diagnosis and treatment of patients who present with respiratory compromise are challenging. What happens when these patients do not respond to your intervention, and their condition declines rapidly? Having a variety of differential diagnoses is key. An addition to your differential list can include a rare disorder of non-Langerhans cells histiocytosis also known as Erdheim-Chester disease. This disease often presents as an interstitial lung disease that fails many different treatment modalities. A full understanding of how this disease process works is still being investigated. Provided are a literature review and case study for better understanding of this disease.	21654219
2011 May 18	Thorac Cardiovasc Surg	Erdheim-Chester Disease in a Female Cardiac Surgery Patient	Mahoozi HR, Zittermann A, Hakim Meibodi K, Burchert W, Gummert JF, Mirow N.	SourceClinic for Thoracic and Cardiovascular Surgery, Heart Center NRW, Bad Oeynhausen, Germany	We report a case of Erdheim-Chester disease (ECD) with isolated cardiac involvement in a 74-year-old female patient. The patient initially presented with superior vena cava syndrome and PET-CT imaging demonstrating an obstructing hypermetabolic lesion in the right atrium, and a distinct nonobstructing hypermetabolic lesion in the left atrium, expected to be malignant. There was no evidence of extracardiac disease. At surgical exploration, consistent with malignancy, the right atrial tumor was found to have grown into the pericardium and was resected to address symptoms and for histological diagnosis which revealed ECD on immunohistochemistry. We conclude that isolated cardiac ECD should be included in the surgical strategy for cardiac tumors showing infiltrative growth.	21594819
2011 Mar	Zhonghua Er Ke Za Zhi	Erdheim-Chester disease in a child: case report	Wen C, Liang QC, Wan WQ		[Article in Chinese]	21575377
2011 Jun	Pathology	Erdheim-Chester disease with extensive coronary arterial involvement	Vaideeswar P, Vaz WF.	Departments of Pathology (Cardiovascular and Thoracic Division), India †Forensic Medicine, Seth G. S. Medical College and K. E. M. Hospital, Mumbai, India		21566495
2011 Apr 6	Cen Eur Neurosurg	Erdheim-Chester Disease - A Rare Differential Diagnosis of Eosinophilic Granuloma. A Case Report	Platz R, Romeike BF, Pandey DK, Kalff R, Reichart R.	University Clinic Jena, Neurosurgical Clinic, Jena, Germany.		21472656
2011 Apr	J Bone Joint Surg Am	Total knee arthroplasty in a patient with erdheim-chester disease with massive joint destruction: a case report	Steinert AF, Reppenhagen S, Baumann B, Rudert M, Nöth U.	Department of Orthopaedic Surgery, König-Ludwig-Haus, Julius-Maximilians-University Würzburg, Brettreichstrasse 11, D-97074 Würzburg, Germany. a-steinert.klh@uni-wuerzburg.de		21471409

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Mar	Intern Med J.	Erdheim-Chester disease presenting with large pericardial effusion: a mimic of disseminated malignancy	Jain P, Jepson N, Lawford R.	Department of Cardiology Prince of Wales Hospital, Sydney, New South Wales, Australia.		21426470
2011 Mar 21	J Clin Oncol	Scintigraphic Diagnosis of Erdheim-Chester Disease	Balink H, Hemmelder MH, de Graaf W, Grond J.	Medical Centre Leeuwarden, Leeuwarden, the Netherlands hans.balink@znb.nl		21422416
2011	Respiratory Medicine CME	Pulmonary Erdheim-Chester disease: A response to predonisolone	Kumi Yoneda Nagahama, Takuo Hayashi, Tetsutaro Nagaoka, Ryota Kanemaru, Shinsaku Togo, Toshio Kumasaka, Toshimasa Uekusa, Kuniaki Seyama, Kazuhisa Takahashi	Department of Respiratory Medicine, Juntendo University School of Medicine; 2-1-1 Hongo, Bunkyo-ku, Tokyo 113-8421, Japan e-mail: k-yoneda@juntendo.ac.jp	Erdheim-Chester disease (ECD) is a rare non-Langerhan's cell histiocytosis of unknown origin, involving multiple organs. The patient with ECD described here is a 38-year-old man who was admitted to the hospital with dyspnea on exertion. His chest radiograph revealed a diffuse reticulonodular shadow. After the video-assisted thoracoscopic surgery was performed, he was diagnosed as having ECD. A brown eruption on his left temple, when tested by skin biopsy, proved to be ECD. No lesions other than these on the lung and skin were identified, and oral administration of predonisolone successfully treated both of them. Although recovery has followed the administration of predonisolone and chemotherapy for several patients with pulmonary ECD, this is the first report that predonisolone alone provided clinical and objective recovery from pulmonary ECD. This outcome indicates that, of all the many treatments tried for ECD, steroids may become the first-line therapy for pulmonary involvement.	none
2011 Mar 10	Blood	Erdheim-Chester: beyond the lesion	Allen CE, McClain KL	Baylor College of Medicine; Texas Children's Cancer Center; Feigin Center; 1102 Bates St; Houston, TX 77030 e-mail: ceallen@txccc.org	Erdheim-Chester Disease (ECD) is an extraordinarily rare, poorly understood, and often fatal histiocytic disorder. In this issue of Blood, Arnaud and colleagues describe cytokine profiles in serum from patients with ECD and identify a systemic proinflammatory cytokine ECD-signature.	21393495
2011 Feb 24	Am J Dermatopathol	Erdheim-Chester Disease: A Histiocytic Disorder More Than Skin Deep	Skinner M, Briant M, Morgan MB	Michael Morgan, 12901 Bruce B. Downs Blvd., Department of Pathology, University of South Florida, Tampa, Florida 33612	Erdheim-Chester disease is a rare potentially malignant systemic non-Langerhans cell histiocytosis. Although classically described in the pulmonary system and long bones, cutaneous involvement has been chronicled in 2 previous case reports. Herein, we describe a single systemic case afflicting an elderly man with synchronous multifocal cutaneous disease. The previous literature and pertinent differential diagnosis will be discussed.	21358382

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Feb 14	J Neuroophthalmol.	Presence of Erdheim-Chester Disease and Langerhans Cell Histiocytosis in the Same Patient: A Report of 2 Cases.	Pineles SL, Liu GT, Acebes X, Arruga J, Nasta S, Glaser R, Pramick M, Fogt F, Roux PL, Gausas RE.	Division of Neuro-Ophthalmology, Department of Neurology (SLP, GTL), Departments of Ophthalmology (SLP, GTL, REG), Hematology-Oncology (SN), Cardiology (RG), Pathology (FF), and Neurosurgery (PLR), Hospital of the University of Pennsylvania, Philadelphia, Pennsylvania; Departments of Neurology and Ophthalmology (XA, JA), Bellvitge Hospital, Barcelona, Spain; and Department of Pathology (MP), Pennsylvania Hospital, Philadelphia, Pennsylvania.	The histiocytic disorders Langerhans cell histiocytosis (LCH) and Erdheim-Chester disease (ECD), can both present with multisystem involvement, with the central nervous system and the bone, skin, neuroendocrine, cardiac, respiratory, and gastrointestinal systems potentially affected. The 2 entities occasionally can be difficult to distinguish. Both rarely affect the orbit and the central nervous system, and although there are rare reports of patients with coexistent LCH and ECD, there are no reported cases of the 2 diseases that involve both the orbital and neuroendocrine systems. We report 2 such cases, and review the literature of cases of LCH and ECD occurring in the same patient. The presentation of LCH and ECD in certain patients suggests a possible abnormality in the common CD34 progenitor cell. The coexistence of the 2 disease states should be suspected in patients with atypical presentations of either disorder.	21326112

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2011 Jan 14	Blood	CNS involvement and treatment with interferon-alpha are independent prognostic factors in Erdheim-Chester disease: a multicenter survival analysis of 53 patients	Arnaud L, Hervier B, Néel A, Hamidou MA, Kahn JE, Wechsler B, Pérez-Pastor G, Blomberg B, Fuzibet JG, Dubourguet F, Marinho A, Magnette C, Noel V, Pavic M, Casper J, Beucher AB, Costedoat-Chalumeau N, Aaron L, Salvatierra J, Graux C, Cacoub P, Delcey V, Dechant C, Bindi P, Herbaut C, Graziani G, Amoura Z, Haroche J.	Dr Julien Haroche Service de Médecine Interne 2. Groupe Hospitalier Pitié-Salpêtrière, 47-83 bd de l'Hôpital, 75013, Paris, France. Phone number: (33) 1 42 17 80 40 Fax number: (33) 1 42 17 80 44 Email : julien.haroche@psl.aphp.fr	Erdheim-Chester disease (ECD) is a rare form of non-Langerhans histiocytosis, with non-codified therapeutic management and high-mortality. No treatment has yet been shown to improve survival in these patients. We conducted a multicenter prospective observational cohort study to assess whether extraskeletal manifestations and interferon-alpha treatment would influence survival in a large cohort of ECD patients. To achieve this goal, we thoroughly analyzed the clinical presentation of 53 patients with biopsy-proven ECD, and performed a survival analysis using Cox proportional hazard model. Fifty-three patients (39 men and 14 women) with biopsy-proven ECD were followed-up between Nov. 1981 and Nov. 2010. Forty-six patients (87%) received interferon-alpha and/or PEGylated interferon-alpha. Multivariate survival analysis using Cox proportional hazard model revealed that CNS involvement was an independent predictor of death (Hazard Ratio, HR: 2.51, CI95%: 1.28-5.52; p=0.006) in our cohort. Conversely, treatment with interferon-alpha was identified as an independent predictor of survival (HR: 0.32, CI95%: 0.14-0.70; p=0.006). Although definitive confirmation would require a randomized controlled trial, these results suggest that interferon-alpha improves survival in ECD patients. This may be seen as a significant advance, as it is the first time a treatment is shown to improve survival in this multisystemic disease with high mortality.	21239701
2011 Jan 4	Blood	Systemic perturbation of cytokine and chemokine networks in Erdheim-Chester disease: a single-center series of 37 patients.	Arnaud L, Gorochov G, Charlotte F, Lvovschi V, Parizot C, Larsen M, Ghillani-Dalbin P, Hervier B, Kahn JE, Deback C, Musset L, Amoura Z, Haroche J.	Departments of Internal Medicine, Pathology, Immunochemistry & Virology, Hopital Pitie-Salpetriere, AP-HP, Paris, France	Immunopathogenesis of Erdheim-Chester disease (ECD), a rare non-Langerhans cell histiocytosis, is poorly known. In previous studies, various cytokines were detected in ECD lesions, presumably orchestrating lesional histiocyte recruitment. Since ECD lesions are frequently associated with systemic symptoms, we postulated that underlying global immune perturbations might also be revealed. We quantitatively analyzed 23 cytokines in serum samples obtained from a large single-center cohort of 37 ECD patients, and studied the impact of treatment over cytokine production. Interleukin (IL)-6, IL-12, interferon-alpha (IFN-α) and monocyte chemotactic protein-1 (MCP-1) levels were significantly higher in untreated patients than in controls, while interferon-γ inducible protein-10 (IP-10), IL-12, MCP-1, IL-1 receptor antagonist (IL1-RA) were found significantly increased in interferon-alpha treated patients. A biomathematical approach was used to rationalize multiparameter data, in order to generate new hypotheses and identify global control pathways. Interestingly, cytokine profiles proved to be particularly stable at the individual level, and an "ECD signature" further distinguished patients from controls, based on their production of IFN-α, IL-12, MCP-1, IL-4 & IL-7. Altogether, our data underline the systemic immune Th-1 oriented perturbation associated with this condition, and provide clues for the choice of more focused therapeutic agents in this rare disease with non-codified therapeutic management.	21205927

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2010 Nov	Ann Dermatol	Erdheim-chester disease.	Kim MS, Kim CH, Choi SJ, Won CH, Chang SE, Lee MW, Choi JH, Moon KC.	Mi-Woo Lee, M.D., Department of Dermatology, Asan Medical Center, University of Ulsan College of Medicine, 86 Asanbyeonwon-gil, Songpa-gu, Seoul 138-736, Korea. Tel: 82-2-3010-3460, Fax: 82-2-486-7831, Email: miumiu@amc.seoul.kr	Erdheim-Chester disease (ECD) is a rare, non-Langerhans cell histiocytosis of unknown etiology, characterized by multi-organ involvement. ECD is usually diagnosed on the basis of characteristic radiologic and histopathological findings. Lesions may be skeletal and/or extraskeletal in location, and may include the skin, lung, heart, and central nervous system. We describe here a 68-year-old man with multiple yellowish plaques and a pinkish nodule on his face and scalp. He had been previously diagnosed with diabetes insipidus, and recently complained of coughing and dyspnea. Imaging studies showed multiple osteosclerotic lesions of the bones, a moderate amount of pericardial effusion, and multifocal infiltrative lesions in the perirenal space. Histopathological examination of the skin lesions revealed dermal infiltration of foamy histiocytes with multinuclear giant cells. Moreover, laparoscopic biopsy of the perirenal tissue revealed fibrosis with infiltrating foamy histiocytes being CD68-positive and S100-negative. Based on these findings, he was diagnosed with ECD with extraskeletal manifestations, and treated with interferon-alpha.	21165216
2010 Dec 12	J Cutan Pathol	Erdheim-Chester disease presenting with cutaneous involvement: a case report and literature review.	Volpicelli ER, Doyle L, Annes JP, Murray MF, Jacobsen E, Murphy GF, Saavedra AP	Arturo P. Saavedra, MD, PhD, Department of Dermatology, Brigham and Women's Hospital, Harvard Medical School, 221 Longwood Avenue, Boston, MA 02115, USA Tel: +1 617 732 4918 begin_of_the_skype_highlighting +1 617 732 4918 end_of_the_skype_highlighting Fax: +1 617 582 6060 e-mail: asoavedra@partners.org	Erdheim–Chester disease (ECD) is a rare, systemic, non-familial histiocytic disorder, first described by Jakob Erdheim and William Chester in 1930. Most patients have multiple sites of involvement at presentation. The most common site of involvement is the long bones of the axial skeleton, which is seen almost universally, followed by the nervous system, heart, lungs, orbit and retroperitoneum, which are seen in up to 50% of cases.1 Cutaneous involvement is rarely a presenting symptom of ECD, with two reported cases in the English literature.2 The diagnosis of ECD is rarely made by skin biopsy because of the relative rarity of cutaneous involvement as a presenting feature, and also perhaps because of the difficulty in distinguishing the histopathological appearance from potential mimics. The importance of distinguishing ECD from other cutaneous disorders with similar pathology lies in the implications for both treatment and prognosis. ECD is an aggressive, often fatal disorder, with death from disease occurring in greater than 50% of patients.	21143617
2010 Dec 7	Rheumatol Int	FDG-PET in the Erdheim-Chester disease: its diagnostic and follow-up role.	Steňová E, Steňo B, Povinec P, Ondriaš F, Rampalová J.	1st Department of Internal Medicine, Comenius University in Bratislava, Bratislava, Slovakia, stenova@faneba.sk.	A 62-year-old man presented with diabetes insipidus, pulmonary fibrosis, right atrial tumor and bilateral knee osteoarthritis with cystic lesions of distal femur and proximal tibia. Scintigraphy and histological examination of right femur bone biopsy revealed changes characterized for Paget's disease. Re-evaluation of the computer tomography (CT) scans and histological samples revealed diffuse infiltrates of foamy histiocytes in the bone marrow what was consistent with Erdheim-Chester disease. Positron emission tomography/computed tomography (PET/CT) was performed to access the activity and extent of disease.	21136259
2010 Nov	Journal of Clinical Oncology	Response of Histiocytoses to Imatinib Mesylate: Fire to Ashes	Filip Janku, Hesham M. Amin, Dan Yang, Ignacio Garrido-Laguna, Jonathan C. Trent, and Razelle Kurzrock	The University of Texas M. D. Anderson Cancer Center, Houston, TX	No abstract included. Final statement of paper states, “LCH and ECD are rare and often difficult to treat disorders. Our observation suggests that imatinib may be an effective treatment option for some patients with these diseases.”	

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2010 Nov	J Neurol Neurosurg Psychiatry	CP3 Progressive ataxia with unusual radiographic findings	Marshall C, Giovannoni G	drcharlesmarshall@gmail.com	A 72-year-old woman presented with a 2-year history of slowly progressive ataxia, accompanied by 13 kg weight loss. MRI imaging of the brain revealed T2-weighted hyperintensities in the pons, midbrain and cerebellum. Multiple blood and CSF investigations for metabolic, inflammatory, infective and genetic aetiologies were negative or normal. A paraneoplastic cause was sought. CT revealed circumferential sheathing of the thoracic and abdominal aorta by abnormal soft tissue. The right kidney was enlarged, with perinephric enhancement and stranding. There were sclerotic bone lesions in multiple ribs, the left humeral midshaft, and both iliac blades. Coarsening of the trabecular pattern was seen in the proximal femurs. Technetium bone scanning showed increased uptake in the sclerotic lesions, and in a symmetrical diametaphyseal pattern in the distal femurs and proximal tibiae. These imaging findings were recognised to be pathognomonic for Erdheim Chester disease, a rare non-Langerhans histiocytosis. The diagnosis was supported by biopsy of the periaortic soft tissue. Progressive ataxia has been described in Erdheim Chester due to infiltration of the cerebellum by histiocytes, but never as the presenting feature. This patient went on to develop other manifestations of histiocytosis, including pituitary, cardiac and pulmonary involvement. Treatment with interferon-α was initiated.	20972058
2010	Respiratory Medicine CME (2010)	Pulmonary Erdheim-Chester disease: A response to predonisolone	Kumi Yoneda Nagahama, Takuo Hayashi, Tetsutaro Nagaoka, Ryota Kanemaru, Shinsaku Togo, Toshio Kumasaka, Toshimasa Uekusa, Kuniaki Seyama, Kazuhisa Takahashi	Department of Respiratory Medicine, Juntendo University School of Medicine; 2-1-1 Hongo, Bunkyo-ku, Tokyo 113-8421, Japan, k-yoneda@juntendo.ac.jp	Erdheim-Chester disease (ECD) is a rare non-Langerhan's cell histiocytosis of unknown origin, involving multiple organs. The patient with ECD described here is a 38-year-old man who was admitted to the hospital with dyspnea on exertion. His chest radiograph revealed a diffuse reticulonodular shadow. After the video-assisted thoracoscopic surgery was performed, he was diagnosed as having ECD. A brown eruption on his left temple, when tested by skin biopsy, proved to be ECD. No lesions other than these on the lung and skin were identified, and oral administration of predonisolone successfully treated both of them. Although recovery has followed the administration of predonisolone and chemotherapy for several patients with pulmonary ECD, this is the first report that predonisolone alone provided clinical and objective recovery from pulmonary ECD. This outcome indicates that, of all the many treatments tried for ECD, steroids may become the first-line therapy for pulmonary involvement.	
2010 Jul-Aug	Tumori	Polyostotic sclerosing histiocytosis (Erdheim-Chester disease) treated with combined vertebroplasty and radiation therapy	Franco P, Filippi AR, Ciammella P, Botticella A, Namysl-Kaletka A, Ricardi U.	Department of Medical and Surgical Sciences, Radiation Oncology Unit, University of Torino, Ospedale S. Giovanni Battista, Turin, Italy. pierfrancesco.franco@gmail.com	Erdheim-Chester disease is an uncommon form of non-Langherans-cell histiocytosis, with a heterogeneous range of systemic manifestations and a pattern of typical clinico-pathological and radiological features. Symmetric sclerotic radiological alterations of the long bones are peculiar, such as the infiltration of several organs by lipid-laden histiocytes. Radiation therapy has been anecdotally employed in a palliative setting in order to relieve symptoms mainly due to cerebral, retro-orbital and skeletal localizations. Exclusive osseous involvement is rarely described in the medical literature. Moreover, the role, timing and schedule of radiotherapy in this subset of patients remain controversial. We herein report on a case of osseous-only Erdheim-Chester disease treated with a combined modality approach including transoral vertebroplasty and external beam radiation therapy, which gave an analgesic effect that lasted 1 year, with no treatment-related side effects.	20968148

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2010 Oct 15	J Neuroradiol	Central nervous system involvement in systemic diseases: Spectrum of MRI findings [Article in French]	Drier A, Bonneville F, Haroche J, Amoura Z, Dormont D, Chiras J.	Service de neuroradiologie, hôpital Pitié-Salpêtrière, 47-83, boulevard de l'Hôpital, 75013 Paris, France	Central nervous system (CNS) involvement in systemic disease (SD) is unusual. MRI features of such lesions are unfamiliar to most radiologists. The diagnosis of SD is still based on clinical features and laboratory findings but some characteristic MRI findings exist for each SD: micronodular leptomeningeal enhancement in sarcoidosis, diffuse or focal pachymeningeal involvement in Wegener disease, dentate nuclei and brain stem lesions in Langerhans cell histiocytosis, meningeal masses, dentate nuclei lesions and periarterial infiltration in Erdheim-Chester disease, meningeal masses in Rosai-Dorfman disease, veinular pontic lesions and cerebral vein thrombosis in Behçet, supratentorial microvascular lesions in lupus and antiphospholipid and Gougerot-Sjögren syndrome. In this work, we explain, describe and illustrate the most characteristic MRI findings for each disease.	20952066
2010 Oct 15	Clin Rheumatol	Erdheim-Chester disease.	Akkara Veetil BM, Luthra H.	Division of Rheumatology, Mayo Clinic, 200 First St. SW, Rochester, MN, 55905, USA, bharathmanu@yahoo.co.in	Erdheim-Chester disease is characterized by long bone pain and symmetric sclerosis of the diametaphyseal portions of the long bones. It is an important differential diagnosis of sclerotic disease of the bones.	20949296
2010 Sep 13	J Clin Neurosci	Surgical treatment of intracranial Erdheim-Chester disease	Alfieri A, Gazzeri R, Galarza M, Neroni M	Department of Neurological Surgery, Martin-Luther University Halle-Wittenberg, Ernst-Grube Straβe 40, Halle, Germany	We review the clinical presentation, radiological and histological characteristics, and the natural history, of intracranial Erdheim-Chester disease (ECD). ECD is a rare form of non-Langerhans histiocytosis that affects multiple organs. It is clinically characterized by leg pain, exophthalmos and diabetes insipidus (DI). Central nervous system involvement is rare, with only 27 patients reported in the international literature. DI and cerebellar signs represent the most common neurological symptoms. Its treatment is controversial. Intracranial surgical procedures for ECD have been reported in 11 patients with a complete surgical resection performed in six, and an intracerebral biopsy performed in five patients. In seven patients the cranial procedures represented the initial diagnostic method. Surgical resection and radiation therapy have been used in the further management of these cerebral lesions.	20843693
2010 Jul-Aug	J Radiol	[Erdheim-Chester disease with mesenteric involvement.] [Article in French]	Lamboley J, Le Moigne F, Felten D, Crozes C, Farthouat P, Pavic M.	Service d'Imagerie médicale, Hôpital d'instruction des armées Desgenettes, 108, boulevard Pinel, 69275 Lyon cedex 03.		20814365
2010 Sep	J Rheumatol	Erdheim-Chester disease	Lee Y, Pearce D.	St. Michael's Hospital - Medicine, 30 Bond St., Toronto, Ontario M5B 1W8, Canada. leeyuna@smh.toronto.on.ca		20810522

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2010 Aug 19	Blood	Rationale and efficacy of interleukin-1 targeting in Erdheim-Chester disease	Aouba A, Georgin-Lavialle S, Pagnoux C, Martin Silva N, Renand A, Galateau-Salle F, Le Toquin S, Bensadoun H, Larousserie F, Silvera S, Provost N, Candon S, Seror R, de Menthon M, Hermine O, Guillevin L, Bienvenu B.	Department of Internal Medicine, French National Referral Center for Langerhans cell histiocytosis, Hopital Cochin, Universite de Paris Descartes, Assistance Publique-Hopitaux de Paris, France	Erdheim-Chester's disease (ECD) pathophysiology remains largely unknown. Its treatment is not codified and usually disappointing. Interferon-alpha (IFN-alpha) therapy lacks efficacy for some life-threatening manifestations and has a poor tolerance profile. Because IL1Ra synthesis is naturally induced after stimulation by IFN-alpha, we hypothesized that recombinant IL1Ra (anakinra) might have some efficacy in ECD. We treated 2 patients who had poor tolerance or contraindication to IFN-alpha with anakinra as a rescue therapy and measured their serum C-reactive protein (CRP), interleukin-(IL)-1beta, IL-6, and monocytic membranous IL-1alpha (mIL-1alpha) levels prior, under and after therapy. Another untreated ECD patient and 5 healthy subjects were taken as controls. After treatment, fever and bone pains rapidly disappeared in both patients, as well as eyelid involvement in one patient. In addition retroperitoneal fibrosis completely or partially regressed and CRP, IL-6 and mIL-1alpha levels decreased to within the normal and controls range. Beside injection site reactions, no adverse event was reported. Therefore, our results support a central role of the IL1 network, which appeared to be overstimulated in ECD. Its specific blockade using anakinra thereby opens new pathophysiology and therapeutic perspectives in ECD.	20724540
2010 Jul 26	Arthritis Rheum	Pulmonary involvement in Erdheim-Chester disease: A single-center experience of 34 patients and review of the literature	Arnaud L, Pierre I, Beigelman C, Capron F, Brun AL, Rigolet A, Girerd X, Weber N, Piette JC, Grenier PA, Amoura Z, Haroche J.	Department of Internal Medicine 2, Pitié-Salpêtrière Hospital, UPMC University Paris 6, AP-HP, 47-83 bd de l'Hôpital, 75013, Paris, France.	OBJECTIVE: Erdheim-Chester disease (ECD) is a rare form of non-Langerhans' cell histiocytosis that may present with pulmonary involvement. This study was undertaken to evaluate the characteristic features of pulmonary involvement of ECD, in the largest single-center series of patients reported to now. METHODS: We performed a retrospective study of the characteristics of 34 consecutive biopsy-proven ECD patients referred to the internal medicine department of Pitié-Salpêtrière Hospital (Paris, France) between 1981 and Nov. 2008. RESULTS: Data were obtained from 23 men and 11 women. Median age at diagnosis was 53.7 yrs (range: 16-73 yrs) and median follow-up was 3.5 years (1.4-5.3 yrs). Eight patients (26%) had pulmonary symptoms. High-resolution chest CT (HRCT)-scan revealed involvement of lung parenchyma in 18 patients (53%), and of the pleura in 14 patients (41%). The bronchoalveolar lavage fluid analysis revealed presence of an opalescent aspirate in all patients studied. Treatment with corticosteroids and/or interferon-alpha resulted in a marked improvement of the pulmonary lesions only in a single patient. Comparison of survival between patients with and without pulmonary involvement yielded no significant difference (p=0.82). CONCLUSION: Pulmonary involvement of ECD has been overlooked in previous reports. HRCT reveals typical lesions in most patients. There is no clear response of these lesions to corticosteroids and interferon alpha. The overall prognosis of the disease is poor, but pulmonary involvement does not appear to be a major prognostic factor in ECD.	20662053

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Jul 23	Presse Med.	Erdheim-Chester disease: Radiological findings	Versini M, Jeandel PY, Fuzibet JG, Ianessi A, Hauger O, Amoretti N.	CHU de Nice, service de médecine interne, 06202 Nice cedex 3, France.	CONTEXT: Erdheim-Chester disease is a rare non-Langerhans form of histiocytosis. For the past years, the disease has been reported with an increasing frequency, linked to a better knowledge of it's radiological pattern. Indead, it shows specific imaging appearances, that should be recognized. METHODS: We report four cases illustrating those typical imaging findings. RESULTS: Common X-rays films show bilateral and symmetric heterogeneous osteosclerosis of the metaphysis and the diaphysis in the lower limbs long bones, with paget's disease-like pattern. Magnetic resonance imaging depicts a replacement of the normal fatty bone marrow by a heterogeneous high intensity signal infiltrate on T1 fat-suppressed weighted imaging with intravenous injection of gadolinium and T2 fat-suppressed weighted sequences, sparing the subchondral bone. Bone scintigraphy reveals a pathognomonic bilateral and symmetric increased uptake affecting both diaphysis and metaphysis of the femur and the tibiae. Tomodensitometry enable to disclose visceral and vascular involvement, showing typical "hairy kidney" appearance and perivascular infitration. CONCLUSION: Erdheim-Chester disease may be a life-threatening disease. A good knowledge of it specific imaging features seems to be crucial for early management and improved prognosis. (Article in French)	20656448
20633799	2010 Aug	Combined Erdheim-Chester disease and Langerhans cell histiocytosis of skin are both monoclonal: a rare case with human androgen-receptor gene analysis.	Tsai JW, Tsou JH, Hung LY, Wu HB, Chang KC	Department of Anatomic Pathology, E-Da Hospital, Kaohsiung County, Taiwan.	BACKGROUND: Erdheim-Chester disease (ECD) is a rare xanthogranulomatous histiocytic disorder. Langerhans cell histiocytosis (LCH) is a proliferative disorder of histiocytes with a phenotype similar to dendritic Langerhans cells. Both are derived from myeloid stem cells in the bone marrow and, thus, can overlap. OBJECTIVE: We report a rare case of hybrid LCH and ECD of the skin with systemic ECD. METHODS: Pathologic examinations and human androgen-receptor gene assay were used to study this case. RESULTS: A 34-year-old woman presented with recurrent ulcerative skin lesions on both thighs associated with polydipsia and polyuria since childhood. Radiography revealed osteosclerosis of bilateral distal tibias and soft tissue masses of bilateral chest walls and ankles. Pathologically, the chest wall lesions showed dense aggregates of lipid-laden histiocytes, which were CD68(+)/CD163(+)/S100(-)/CD1a(-). Combined with the clinical and radiographic findings, this xanthogranulomatous infiltrate was consistent with ECD. However, thigh skin showed discrete foci of a xanthogranulomatous infiltrate and S100(+)/CD1a(+) Langerhans cells with eosinophils. In addition, Birbeck granules were found. Dissected tissues from both ECD and LCH were monoclonal, supporting their neoplastic nature. LIMITATIONS: Single case report is a limitation. CONCLUSION: ECD and LCH may have a close association with divergent differentiation from the same stem cells under different microenvironmental conditions.	20633799
2010 Jun 29	Pediatr Blood Cancer	Chemotherapy and interferon-alpha treatment of Erdheim-chester disease.	Jeon IS, Lee SS, Lee MK.	Department of Pediatrics, Gil Medical Center, Gachon Medical School, Gachon University of Medicine and Science, Incheon, Korea.	Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis of an unknown origin. The prognosis of ECD is variable, and it mainly depends on the involved anatomic sites. The treatment modalities have not been standardized. Interferon-alpha (IFN) has been reported to be effective in the management of ECD. We report here on an uncommon case with ECD in a 17-year-old female who had multiple lesions in the whole body and she was treated with chemotherapy and IFN. She has remained disease-free for 2 years after the completion of treatment. Pediatr Blood Cancer (c) 2010 Wiley-Liss, Inc.	20589628

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Jun 20	Eur Radiol.	Erdheim-Chester disease: CT findings of thoracic involvement.	Brun AL, Touitou-Gottenberg D, Haroche J, Toledano D, Cluzel P, Beigelman-Aubry C, Piette JC, Amoura Z, Grenier PA.	Radiologie, Hôpital Pitié-Salpêtrière, Assistance Publique-Hôpitaux de Paris, Université Pierre et Marie Curie, Paris VI, Paris, France.	OBJECTIVE: To retrospectively assess the association of mediastinal, cardiovascular and pleuropulmonary findings on chest CT of 40 patients with immunohistochemically and histologically proven Erdheim-Chester disease (ECD). METHODS: The multidetector chest CT images of 40 ECD patients were reviewed in consensus by chest and cardiovascular radiologists. RESULTS: Thirty-four (85%) patients had periaortic infiltration that extended around the aortic branches of 29 (73%). Perivascular infiltration extended into the cardiac sulci in 22 (55%) ($p < 0.005$). Infiltration involved the right atrium wall in 12 patients, associated with severe narrowing of the atrial lumen in 8. Pericardial effusion and/or thickening were observed in 24 (60%) patients. Lung involvement, seen in 22 (55%) patients, was associated with mediastinal infiltration (20; $p < 0.005$) and pleural thickening or effusion (16; $p = 0.001$); it consisted of smooth interlobular septa (21), subpleural thickening (13), poorly defined centrilobular nodular opacities (9), ground-glass opacities (8) and/or lung cysts (5). CONCLUSION: The detailed description of thoracic ECD involvement seen in these patients showed that infiltration into the mediastinal spaces including the pericardium, coronary sulci and right atrium is frequently associated with pleural and interstitial lung diseases.	20563815
2010 Jun 15.	Intern Med.	A case of osteoarthropathy due to erdheim-chester disease with overlapping Langerhans' cell infiltration.	Naruse H, Shoda H, Okamoto A, Oka T, Yamamoto K.	Department of Allergy and Rheumatology, Graduate School of Medicine, the University of Tokyo, Tokyo, Japan.	Histiocytosis sometimes involves the joints, and is one of the important differential diagnoses of osteoarthropathy. A 31-year-old man presented with recurrent fever and bilateral knee arthritis two years prior to admission. He also showed the hypopituitary mass lesion and partial hypopituitarism. X-ray studies showed both osteosclerotic and osteolytic lesions near the large joints. Histological findings of bone biopsy revealed foamy macrophage infiltration, which were CD68+CD1a-S100-, and Erdheim-Chester disease was diagnosed. In addition, CD68+CD1a+ Langerhans' cells also aggregated in the same lesions, and we thought this case was a rare variant of Erdheim-Chester disease with overlapping histiocytic invasion.	20558949
2010 Jun	Br J Neurosurg.	Erdheim-Chester disease mimicking multiple meningiomas	Donaldson G, Bullock P, Monson JP.	The London Clinic Centre for Neurosurgery, London, UK. georgedon68@hotmail.co.uk	Erdheim-Chester disease is a rare non-Langerhans cell histiocytosis with systemic manifestations. We present a case report of a patient initially diagnosed with multiple meningiomas, wherein an orbital biopsy disclosed the true nature of the disease.	20465459
2010 May	Acta Neurochir	Meningioma-like lesions in Erdheim Chester disease.	Naqi R, Azeemuddin M, Idrees R, Wasay M.	Department of Radiology, Aga Khan University, Karachi, Pakistan		20449616

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010	RSNA	Cerebral, Facial, and Orbital Involvement in Erdheim-Chester Disease: CT and MR Imaging Findings	Aurélie Drier, MD, Julien Haroche, MD, PhD, Julien Savatovsky, MD, Gaelle Godenèche, MD, Didier Dormont, MD, Jacques Chiras, MD, Zahir Amoura, MD and Fabrice Bonneville, MD, PhD	aureliedrier@gmail.com	Purpose: To retrospectively review the brain magnetic resonance (MR) imaging and computed tomographic (CT) findings in patients with Erdheim-Chester disease (ECD). Materials and Methods: The ethics committee required neither institutional review board approval nor informed patient consent for retrospective analyses of the patients' medical records and imaging data. The patients' medical files were retrospectively reviewed in accordance with human subject research protocols. Three neuroradiologists in consensus analyzed the signal intensity, location, size, number, and gadolinium uptake of lesions detected on brain MR images obtained in 33 patients with biopsy-proved ECD. Results: Thirty patients had intracranial, facial bone, and/or orbital involvement, and three had normal neurologic imaging findings. The hypothalamic-pituitary axis was involved in 16 (53%) of the 30 patients, with six (20%) cases of micronodular or nodular masses of the infundibular stalk. Meningeal lesions were observed in seven (23%) patients. Three (10%) patients had bilateral symmetric T2 high signal intensity in the dentate nucleus areas, and five (17%) had multiple intraaxial enhancing masses. Striking intracranial periarterial infiltration was observed in three (10%) patients. Another patient (3%) had a lesion in the lumen of the superior sagittal sinus. Nine (30%) patients had orbital involvement. Twenty-four (80%) patients had osteosclerosis of the facial and/or skull bones. At least two anatomic sites were involved in two-thirds (n = 20) of the patients. Osteosclerosis of the facial bones associated with orbital masses and either meningeal or infundibular stalk masses was seen in eight (27%) patients.	20413768

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Jan-Apr	Hell J Nucl Med.	Erdheim-Chester's disease as a differential diagnosis of "hot" kidneys on bone scintigraphy.	Javadi H, Malek H, Neshandar Asli I, Mogharrabi M, Assadi M.	Department of Nuclear Medicine, 5-Azar Hospital, Golestan University of Medical Science Gorgan, Iran. assadipoya@yahoo.com.	To the Editor: We read with interest a case of Erdheim-Chester disease (ECD) published in HJNM 2008; 10: 164-167 and we would like to present another case which differs from the above as having an unusual bone involvement and "hot" kidneys on bone scintigraphy. The patient was a 46 years old man admitted for evaluation of the pain that he had in his lower limbs for the last 3 years. He also had weakness, weight loss and diabetes insipidus. Physical examination revealed pitting edema of the lower limbs and some cutaneous xanthelasmata. Serum creatinine was normal. Sonography of the kidneys demonstrated increased renal size (145x67x28mm for left kidney and 140x66x24mm for right kidney) and some corticomedullary loss of image differentiation without evidence of obstructive calyceal dilatation. X-rays of both proximal and distal femora showed symmetric metaphyseal and diaphyseal involvement of lesions including mixed osteosclerosis and lytic areas. Bone scintigraphy with technetium-99m-methylene diphosphonate ((99m)Tc-MDP) revealed multiple bone involvement. Sites of symmetrical increased radionuclide uptake included humeri, scapulae, radii, femori, tibiae, tarsal and metatarsal bones. Right iliac bone also showed focal hyperactivity. The skull and the vertebral column were intact. Furthermore, both kidneys demonstrated markedly increased radionuclide uptake. The patient had not taken any nephrotoxic drugs before or during our examination. Bone biopsy from right femoral lateral epicondyle showed fibro-collagenous and fatty tissue infiltrated by clusters of foamy histocytes with central vesicular nuclear and abundant vacuolated cytoplasm. Some touton-shape giant cells were noted. There was also small aggregation of histiocytic like cells with eosinophillic cytoplasm and ovaloid nuclei. Renal biopsy demonstrated similar parenchymal infiltration. These pathologic findings supported the diagnosis of ECD.. Our case demonstrated bilaterally marked renal radionuclide uptake resulting from bilateral renal parenchymal involvement. In addition, there was an unusual asymmetry in size of both kidneys that could be due to different involvement of each kidney. To the best of our knowledge, there are few reported cases of ECD that had significantly high renal parenchymal uptake of the bone imaging agent, althouth nephromegaly and "hairy kidney" appearance on an abdominal CT have been reported. Thus ECD should be considered in the spectrum of differential diagnosis of "hot" kidneys on radionuclide bone scanning. In addition, "hot" kidneys may imply renal parenchymal involvement during disease progression. In conclusion, besides other typical bone scan findings, Erdheim-Chester disease should be considered in the spectrum of differential diagnosis of "hot" kidneys on bone scintigraphy.	20411179
2010 Apr 20	Eur J Echocardiogr	Multimodality imaging showing complete cardiovascular involvement by Erdheim-Chester disease.	Alharthi MS, Calleja A, Panse P, Appleton C, Jaroszewski DE, Tazelaar HD, Mookadam F.	Cardiovascular Division, Internal Medicine Department, Mayo Clinic Arizona, Scottsdale, AZ, USA.	Erdheim-Chester disease (ECD) is a multisystem non-Langerhans form of cell histiocytosis. Histiocytic infiltration leads to xanthogranulomatous infiltrates of multiple organ systems. Erdheim-Chester disease was first reported in1930, only 320 cases reported in the literature. Cardiac involvement in ECD carries worst prognosis beside the central nervous system. We report the first case with pan-cardiac involvement diagnosed with multimodality imaging.	20406735

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Mar 10	Neuropathology	Non-Langerhans cell histiocytosis with isolated CNS involvement: An unusual variant of Erdheim-Chester disease	Conley A, Manjila S, Guan H, Guthikonda M, Kupsky WJ, Mittal S.	Department of Neurosurgery, Karmanos Cancer Institute, Wayne State University, and Detroit Medical Center, Detroit, MI, USA.	Benign histiocytic proliferations are identified by their component cells and classified as either Langerhans cell histiocytosis or non-Langerhans cell histiocytosis. We report a 58-year-old Caucasian woman who presented with diabetes insipidus and was found to harbor a large suprasellar mass. Histopathological analysis was consistent with non-LCH. The differential diagnoses included juvenile xanthogranuloma, adult-onset xanthogranuloma, xanthoma disseminatum, Rosai-Dorfman disease, and Erdheim-Chester disease. Immunohistochemical examination demonstrated a proliferation of large lipid-laden histiocytic cells which were positive for CD68, negative for S100 protein, and showed only faint, background staining for CD1a. We present a case of an autopsy-confirmed non-Langerhans cell histiocytosis limited to the central nervous system and evaluated with both immunohistochemical and ultrastructural studies. Based on the multifocality, anatomic distribution, and immunostaining features, a diagnosis of Erdheim-Chester disease was made. This is only the second reported case of Erdheim-Chester disease with intracranial involvement but absence of extracerebral manifestations. Given the overlapping clinicopathologic, radiographic, and immunohistochemical profiles, differentiating between these rare histiocytic disorders can often present a significant diagnostic challenge. A systematic approach using all available clinical, laboratory, radiographic, histologic, immunohistochemical and ultrastructural data is essential for proper discrimination between the numerous histiocytoses.	20337948

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Feb	Vnitr Lek.	Central diabetes insipidus in adult patients--the first sign of Langerhans cell histiocytosis and Erdheim-Chester disease. Three case studies and literature review			Central diabetes insipidus with an onset in adulthood is very rare. Unlike in children, central diabetes insipidus in adults is more frequently caused by inflammatory processes and neoplastic infiltrations that do not originate from the neuronal tissue than primary neuronal tissue tumours. Rare histiocytic neoplasias (Langerhans cell histiocytosis, xanthogranulomatosis and Erdheim-Chester disease) have a specific affinity to hypothalamus and the pituitary stalk not only in paediatric patients but also when occurring in adults. We describe 3 cases of central diabetes insipidus with an onset in adulthood. Diabetes insipidus was the first sign of Langerhans cell histiocytosis in 2 patients, and it was the first sign of Erdheim-Chester disease in one patient. MR imaging showed pathological infiltration and dilated pituitary stalks in all 3 patients. PET-CT proved useful in differential diagnosis, showing further extracranial pathological changes either on the basis of significant glucose accumulation or on the basis of CT imaging. The Langerhans cell histiocytosis in the first patient has also manifested itself as an infiltration of the perianal area with intensive accumulation of fluorodeoxyglucose (FDG) - SUV 8.6 and gingival inflammation indistinguishable from parodontosis. Histology of the perianal infiltrate confirmed Langerhans cell histiocytosis. Infiltration of the pituitary stalk disappeared from the MR image after 4 cycles of 2-chlordeoxyadenosin (5 mg/m2 5 consecutive days). The PET-CT of the 2nd patient showed only borderline accumulation of FDG in the ENT area, while simultaneously performed CT imaging showed cystic restructuring of the pulmonary parenchyma and nodulations consistent with pulmonary Langerhans cell histiocytosis. Bronchoalveolar lavage identified higher number of CD1 and S100 positive elements, consistent, once again, with pulmonary LCH also affecting pituitary stalk and ear canal. The PET-CT of the third patient showed increased activity in the long bones and ilium near the sacroiliac joint. Biopsy of the focus in the ilium confirmed foam histiocyte infiltration immunochemically corresponding to Erdheim-Chester disease. Additional imaging assessments revealed the presence of further signs of the disease. Pituitary infiltrate biopsy in this patient did not elucidate the diagnosis but resulted in complete panhypopituarism. Central diabetes insipidus in adulthood might be the first sign of so far undiagnosed extracranial disease, in our case of histiocytic neoplasias, and PET-CT has an excellent potential to detect extracranial symptoms of these conditions. Therefore, the high-risk pituitary stalk infiltrate biopsy should always be preceded by comprehensive examination aimed at identification of extracranial manifestations of the pituitary gland diseases.	20329585
2010 Mar	Neth J Med	An X-ray that helps to solve the puzzle.	Bech AP, Reichert LJ.	Department of Internal Medicine, Rijnstate Hospital, Arnhem, the Netherlands. annekebech@hotmail.com		20308710
2010 Mar 20	Med Clin (Barc).	Erdheim-Chester disease with bone lesion and retroperitoneal fibrosis	Rodríguez Avila EE, Rubio Barbón S, Fonseca Aizpuru EM, De La Tassa JM.	Servicio de Medicina Interna, Hospital de Cabueñes, Gijón, Asturias, España.		20307894

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Mar 16	Int Urol Nephrol	Erdheim-Chester disease as cause of end-stage renal failure: a case report and review of the literature	Sanchez JE, Mora C, Macia M, Navarro JF.	Hospital Universitario Central de Asturias, Oviedo, Spain, jesastur@hotmail.com.		20232144
2010 Mar 10	Neurochirurgie	Histiocytic disorders with orbital involvement	Civit T, Colnat-Coulbois S, Marie B.	Département de neurochirurgie, hôpital Central, CHU de Nancy, 29, avenue de Lattre-de-Tassigny, 54000 Nancy, France. t.civit@chu-nancy.fr		20226484
2010 Mar 4	Am J Surg Pathol	Erdheim-Chester Disease Presenting as Bilateral Clinically Malignant Breast Masses	Provenzano E, Barter SJ, Wright PA, Forouhi P, Allibone R, Ellis IO	Departments of *Histopathology daggerRadiology double daggerSurgery, Addenbrookes Hospital and Cambridge Breast Unit, Cambridge UK section signDepartment of Histopathology, Nottingham University Hospitals NHS Trust, Nottingham	Erdheim-Chester disease is a rare non-Langerhans cell histiocytosis of unknown etiology, the commonest sites of involvement being the long bones, skin, orbit, pituitary and retroperitoneum. Breast involvement is rare, with only four reported cases in the English literature. We present a case of a 78-year-old female presenting with bilateral clinically malignant breast masses, with mammographic and ultrasound findings suggestive of locally advanced bilateral breast cancer. Core biopsies from both breasts showed identical features, with a diffuse xanthomatous infiltrate with scattered Touton-type giant cells and a patchy lymphocytic infiltrate. The cells were CD68 positive, and negative for S100, CD1a and a broad panel of cytokeratins. The patient has a background history of cerebrovascular disease with carotid artery stenosis, and subsequently developed rapid restenosis after carotid endarterectomy. With the combined clinical history and classic histological findings in the breast, a diagnosis of Erdheim-Chester disease was made. This is the fifth case report of Erdheim-Chester disease involving the breast, and only the second case with breast lesions as the presenting symptom. Perivascular infiltration is also a rare but recognized presentation of Erdheim-Chester disease. Histiocytic proliferations including ECD can mimic breast carcinoma clinically, radiologically, and histologically, and should be considered in the differential diagnosis of breast mass lesions.	20216377
2010 Mar	J Neurosurg Spine	Atypical spine involvement of Erdheim-Chester disease in an elderly male	Allmendinger AM, Krauthamer AV, Spektor V, Aziz MS, Zablow B	Department of Radiology and Pathology, St. Vincent's Catholic Medical Center, New York, New York 10011, USA. amallmendinger@gmail.com	Erdheim-Chester disease is a rare form of non-Langerhans histiocytosis presenting in the 5th through 7th decades of life. Osseous manifestations include symmetrical sclerosis of the long bones and, rarely, the spine. Central nervous system disease commonly affects the white matter tracts as well as the orbits, but epidural disease is rare. To the best of the authors' knowledge, simultaneous epidural and skeletal spine disease has not been reported. The MR imaging characteristics of skeletal spine disease have also not been reported. The authors describe the case of an 87-year-old man with both epidural and skeletal spine disease. The clinical characteristics, imaging manifestations, and the histological features are discussed.	20192624
2010 Mar	Clin Radiol.	Erdheim-Chester disease presenting with destruction of a metacarpal.	Davies AM, Colley SP, James SL, Sumathi VP, Grimer RJ.	Department of Radiology, Royal Orthopaedic Hospital, Birmingham B31 2AP, United Kingdom.		20152283

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Jan 22	Rheumatology	Erdheim-Chester disease: report on a case and new insights on its immunopathogenesis	Dagna L, Girlanda S, Langheim S, Rizzo N, Bozzolo EP, Sabbadini MG, Ferrarini M.	Marina Ferrarini, Laboratory of Tumor Immunology, Department of Oncology, Scientific Institute H. S. Raffaele, via Olgettina 60, I-20132 Milano, Italy. E-mail: ferrarini.marina@hsr.it		20097905
2009 Dec	Vnitr Lek	Diabetes insipidus followed, after 4 years, with dysarthria and mild right-sided hemiparesis--the first clinical signs of Erdheim-Chester disease. Description and depiction of a case with a review of information on the disease	Adam Z, Balsíková K, Pour L, Krejcí M, Svacina P, Dufek M, Kren L, Hermanová M, Moulis M, Vanícek J, Neubauer J, Mechl M, Prásek J, Staníček J, Koukalová R, Hájek R, Mayer J.	Interní hematologická klinika Lékarské fakulty MU a FN Brno. z.adam@fnbrno.cz	In 2004, diabetes insipidus was the first clinical sign of Erdheim-Chester disease in our patient. Following introduction of substitution therapy with adiuretin, the patient had no further health complaints for four years until 2008 when he gradually developed dysarthria and, consequently, movement disorder in the form of mild right hemiparesis. The first CNS CT scan (2004) did not reveal any pathology. The first pathological MRI of the brain in 2006 - thickening of pituitary stalk by pathological infiltration to 4-5 mm. During the following year, further infiltrates were detected in the CNS. The number and size of CNS infiltrates increased gradually on MRIs performed repeatedly up to 2008. Erdheim-Chester disease has become suspected based on PET-CT examination at the end of 2008. CT showed irregular structure of the skeleton with noticeable sclerotic foci in otherwise osteoporotic bone structure; changes were the most evident in the long bones of lower limbs, in the pelvic bones, skull and arms, while only one vertebra was affected from within the entire spine. Finding ofthickened aortic wall (up to 8 mm) as another pathological circumstance was consistent with the Erdheim-Chester disease-associated changes described as coated aorta. CT scan revealed clear fibrotic changes in the area of retroperitoneum. Applied fluorodeoxyglucose has accumulated in the bone foci described on CTscans as well as in the thickened wall ofthe thoracic and abdominal aorta (SUV 3.6). Tc-pyrophosphonate skeleton scintigraphy showed the same bone foci as PET-CT. Full body MRI showed pathological signal from the bone marrow of the above mentioned locations, particularly during STIR imagining, where there was clear abnormal signal corresponding to accumulated histiocytes, the higher signal of which was well-differentiated from the normal bone marrow. Measurement of bone mineral density with DEXA confirmed reduced density in lumbar vertebrae to the average value of - 2.7 SD (the lowest value was -3.1SD). The disease is associated with elevated inflammatory parameters: leucocytosis, thrombocytosis, elevated CRP and fibrinogen levels. Diagnosis was verified following histological assessment ofiliac bone marrow, where focal infiltrations with foamy histiocytes of typical immunophenotype (CD68+, CD1a-, S100-) were confirmed. Treatment was initiated with chemotherapy consisting of 2g/m2 of cyclophosphamide on day 1 and 200 mg/m2 of etoposide IV infusion on days 1-3, and followed by administration of 5 microg/kg of G-CSF and collection of haematopoietic peripheral blood stem cells (PBSC). PBSC collection was followed by 5-day administration of 5 mg/m2/day of 2-chlorodeoxyadenosine (Litac) administered to the patient at monthly intervals.	20070034

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2010 Jan 5	Acta Neurochirurgica	Neurosurgical biopsy as the initial diagnosis of xanthogranuloma of the Erdheim-Chester disease variety of the infundibulum and optic apparatus: letter to the editor	Abla AA, Wilson DA, Eschbacher JM, Spetzler RF.	Neuroscience Publications; Barrow Neurological Institute, St. Joseph's Hospital and Medical Center, 350 W. Thomas Road, Phoenix, AZ 85013, USA neuropub@chw.edu		20049489
2010 Jan;17	Adv Anat Pathol	Histiocytic disorders of the lung	Nagarjun Rao R, Moran CA, Suster S.	Department of Pathology, Medical College of Wisconsin, Milwaukee, 53226, USA. arao@mcw.edu	Histiocytic proliferations involving the lung span a broad spectrum. Some proliferations are primary; others represent a histiocytic response secondary to conditions in which there may be isolated lung involvement or the lung may be involved as part of a systemic process. Primary histiocytic lung disorders, particularly those of uncertain histogenesis are a heterogeneous and intriguing group of disorders. Although they have been the focus of attention by clinicians and pathologists alike, much is unknown about their etiopathogenesis. Owing to this uncertainty, our understanding of these processes is in a state of flux, and is likely to change as more information is brought to light. This review will focus on pulmonary histiocytic proliferations of uncertain histogenesis. Other histiocytic lesions will be dealt with in brief.	20032634
2009 Dec 15	Neurology	Characteristic brain MRI appearance of erdheim-chester disease.	Bianco F, Iacovelli E, Tinelli E, Locuratolo N, Pauri F, Fattapposta F.	Department of Neurology and ENT, Neuroradiology Unit, University of Rome, "Sapienza," Viale Università 30, 00185 Rome, Italy; federico.bianco@uniroma1.it		20018640
2009 Dec	Arch Pathol Lab Med.	Adult orbital xanthogranulomatous disease: review of the literature.	Guo J, Wang J.	Department of Pathology and Laboratory Medicine, Loma Linda University Medical Center, Loma Linda, California 92354, USA. jguo@llu.edu	This article provides an overview of the pathologic features of adult orbital xanthogranulomatous disease, a rare heterogeneous group of disorders that includes 4 clinical syndromes: adult-onset xanthogranuloma, necrobiotic xanthogranuloma, adult-onset asthma and periocular xanthogranuloma, and Erdheim-Chester disease. The diagnosis is made by biopsy of the lesion, demonstrating tissue infiltration by the hallmarks of xanthoma cells and Touton giant cells. The differential diagnosis is broad, including syndromes within the adult xanthogranulomatous disease category as well as other entities involving the eyelid and the orbital tissues. Because of its rarity and sometimes close similarity to other disease entities, it is often misdiagnosed initially. This article focuses on the morphology and immunohistochemical patterns in diagnosis of adult orbital xanthogranulomatous disease with emphasis on adult-onset asthma and periocular xanthogranuloma in particular, its clinical features and associated systemic manifestations in differential diagnosis, as well as the current management strategy.	19961259

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009	Acta Ophthalmol	Azathioprine and prednisone combination treatment for adult periocular and orbital xanthogranulomatous disease	Ward R. Bijlsma,1 Willem A. van den Bosch,2 Paul L. A. van Daele3 and Dion Paridaens	Ward R. Bijlsma University Medical Centre Utrecht Department of Ophthalmology Heidelberglaan 100 3584 CX Utrecht The Netherlands. Tel: +31 88 755 1683 Fax: + 31 88 755 5417 Email: w.r.bijlsma@umcutrecht.nl	Purpose: To report the authors' experience with azathioprine and prednisone combination for adult periocular and orbital xanthogranulomatous disease. Methods: We identified 13 adults with histology-proven periocular or orbital xanthogranuloma in two tertiary referral orbital centres from 1984 to 2008. Patient records were reviewed and data collected on orbital localization, immune dysfunction, applied treatment and outcome. Results: Five patients with periocular or orbital xanthogranulomatous disease were fully treated with prednisone and azathioprine combination, which resulted in stabilization in two and regression in three. Two other patients had to discontinue azathioprine because of side-effects. Of the non-fully treated prednisone/azathioprine patients, four out of eight progressed. Conclusion: In adult periocular and orbital xanthogranuloma, combined treatment with prednisone and azathioprine yields adequate immunosuppression, often for a prolonged period of time.	
2009 Oct	Best Pract Res Clin Endocrinol Metab.	Pituitary tumours: inflammatory and granulomatous expansive lesions of the pituitary	Carpinteri R, Patelli I, Casanueva FF, Giustina A.	Department of Medical and Surgical Sciences, University of Brescia, Endocrine Service, Montichiari Hospital, via Ciotti 154, 25018 Montichiari, Italy.	Inflammatory and granulomatous diseases of the pituitary are rare causes of sellar masses. Lymphocytic hypophysitis is the most relevant of these disorders, and it is characterised by autoimmune pathogenesis with focal or diffuse inflammatory infiltration and varying degrees of pituitary gland destruction. Endocrine symptoms may include partial or total hypopituitarism, with adrenocorticotropic hormone (ACTH) deficiency being the earliest and most frequent alteration. Pituitary abscess is a rare but potentially life-threatening disease and, in 30-50% of patients, anterior pituitary hormone deficiencies or central diabetes insipidus (DI) at onset may be observed: the earliest manifestation being growth hormone deficiency (GHD), followed by follicle-stimulating hormone (FSH)/luteinising hormone (LH), thyroid-stimulating hormone (TSH) and ACTH deficiencies. Fungal infections of the pituitary are also very rare and include aspergillosis and coccidioidomycosis. Concerning pituitary involvement in systemic diseases, in sarcoidosis endocrine complications are rare, but the hypothalamus and pituitary are the glands most commonly affected. DI is reported in approximately 25-33 % of all neurosarcoidosis cases and is the most frequently observed endocrine disorder. Hyperprolactinaemia and anterior pituitary deficiencies may also occur. Rarely, partial or global anterior pituitary dysfunction may be present also in Wegener's granulomatosis, either at onset or in the course of the disease, resulting in deficiency of one or more of the pituitary axes. Other forms of granulomatous pituitary lesions include idiopathic giant cell granulomatous hypophysitis, Takayasu's disease, Cogan's syndrome and Crohn's disease. The hypotalamic-pituitary system is involved mainly in children with Langerhans' cells histiocytosis who develop DI, which is the most common endocrine manifestation. Anterior pituitary dysfunction is found more rarely and is almost invariably associated with DI. Pituitary involvement may also be observed in another form of systemic hystiocitosis, that is, Erdheim-Chester disease. Tuberculosis is a rare cause of hypophysitis. In conclusion, in patients with a sellar mass and unusual clinical presentation (DI, neurological symptoms), aggressiveness and onset and in the presence of systemic diseases, inflammatory and granulomatous pituitary lesions should be carefully considered in differential diagnosis.	19945028

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009 Nov 14	Radiol Med.	Erdheim-Chester disease: clinical and radiological findings	De Filippo M, Ingegnoli A, Carloni A, Verardo E, Sverzellati N, Onniboni M, Corsi A, Tomassetti S, Mazzei M, Volterrani L, Poletti V, Zompatori M.	Dipartimento di Scienze Cliniche, Sezione di Scienze Radiologiche, Università degli Studi di Parma, Parma, Italy, massimo.defilippo@unipr.it.	PURPOSE: The authors retrospectively reviewed six cases of histologically proven Erdheim-Chester disease (ECD) to evaluate organ involvement and clinical and radiological findings. MATERIALS AND METHODS: Through a search of the pathology databases of four Italian hospitals, we identified six men (mean age, 56 years) with a histological diagnosis of ECD. Histology was performed on retroperitoneal or pulmonary biopsy, depending on disease involvement on imaging. Patients underwent plain radiography of the lower limbs and chest, total-body computed tomography (CT) and bone scintigraphy. Magnetic resonance (MR) imaging was performed in two patients to evaluate the lower limbs and in one patient to study the brain, the chest and the abdomen. RESULTS: Clinical manifestations included dyspnoea (n=2), hydronephrosis (n=2) and bone pain (n=1). Bilateral symmetrical osteosclerosis of the metaphyseal and diaphyseal portions of the lower-limb long bones was present in five patients. Imaging studies revealed extraskeletal manifestations in all patients, including involvement of the retroperitoneal space (n=4), the lung (n=4) and the heart (n=2). CONCLUSIONS: ECD is a multiorgan disease that displays constant involvement of the bones and retroperitoneum; in particular, of the perirenal fat. Although the diagnosis of ECD is histological, imaging can raise suspicion and help to establish a presumptive diagnosis.	19915998
2009 Oct 12	Eur Heart J	Cardiac magnetic resonance characterization of atrial pseudo-mass in Erdheim-Chester disease	Mileto A, Di Bella G, Gaeta M	Department of Radiological Sciences, Policlinico 'G. Martino', University of Messina, Messina, Italy		19825811

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009 Sep 29	Arthritis Rheum.	(18)F-fluorodeoxyglucose-positron emission tomography scanning is more useful in followup than in the initial assessment of patients with Erdheim-Chester disease.	Arnaud L, Malek Z, Archambaud F, Kas A, Toledano D, Drier A, Zeitoun D, Cluzel P, Grenier PA, Chiras J, Piette JC, Amoura Z, Haroche J.	Hôpital Pitié-Salpêtrière, Assistance Publique-Hôpitaux de Paris, and Université Paris 6, Paris, France.	OBJECTIVE: Erdheim-Chester disease (ECD) is a rare form of non-Langerhans' cell histiocytosis. The aim of this study was to assess the value of whole-body scanning with (18)F-fluorodeoxyglucose-positron emission tomography (FDG-PET) in a large cohort of ECD patients from a single center. METHODS: We retrospectively reviewed all PET scans performed on 31 patients with ECD who were referred to our department between 2005 and 2008. PET images were reviewed by 2 independent nuclear medicine specialist physicians and were compared with other imaging modalities performed within 15 days of each PET scan. RESULTS: Thirty-one patients (10 women and 21 men; median age 59.5 years) underwent a total of 65 PET scans. Twenty-three patients (74%) were untreated at the time of the initial PET scan, whereas 30 of the 34 followup PET scans (88%) were performed in patients who were undergoing immunomodulatory therapy. Comparison of the initial and followup PET scans with other imaging modalities revealed that the sensitivity of PET scanning varied greatly among the different organs studied (range 4.3-100%), while the specificity remained high (range 69.2-100%). Followup PET scans were particularly helpful in assessing central nervous system (CNS) involvement, since the PET scan was able to detect an early therapeutic response of CNS lesions, even before magnetic resonance imaging showed a decrease in their size. PET scanning was also very helpful in evaluating the cardiovascular system, which is a major prognostic factor in ECD, by assessing the heart and the entire vascular tree during a single session. CONCLUSION: The results of our large, single-center, retrospective study suggest that the findings of a FDG-PET scan may be interesting in the initial assessment of patients with ECD, but its greater contribution is in followup of these patients.	19790052
2009 Sep	AJR Am J Roentgenol.	AJR teaching file: A rare multisystem disease with distinctive radiologic-pathologic findings	Venkatanarasimha N, Garrido MC, Puckett M, White P.	Department of Radiology, Torbay General Hospital, Lawes Bridge, Torquay, Devon, United Kingdom. nandashettykv@yahoo.com		19696245
2009 Sep 25	Ophthalmologe	Erdheim-Chester disease of the orbit with compressive optic neuropathy.	Manousaridis HK, Casper J, Schittkowski MP, Nizze H, Guthoff RF	Klinik und Poliklinik für Augenheilkunde, Universität Rostock, Doberanerstrasse 140, 18055, Rostock, Deutschland klemanousaridis@kabelmail.de	A 60-year-old man presented with left exophthalmos and deterioration in visual acuity of slow evolution. Bilateral orbital Erdheim-Chester disease was diagnosed. Systemic evaluation revealed a retroperitoneal fibrosis. Treatment with interferon-alpha followed, but bilateral compressive optic neuropathy with visual acuity deterioration and visual field defects evolved. Bilateral orbital decompression was performed.	19777245

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009 Sep 14	J Pediatr Hematol Oncol.	Erdheim-Chester Disease in Childhood: A Challenging Diagnosis and Treatment.	Tran TA, Fabre M, Pariente D, Craiu I, Haroche J, Charlotte F, Eid P, Durrbach A, Taoufik Y, Kone-Paut I.	Department of Pediatrics, Pediatric Rheumatology, Bicêtre University Hospital, Le Kremlin Bicêtre, France	Erdheim-Chester disease is a rare, non-Langerhans systemic histiocytosis characterized by bilateral sclerosis of the metaphyseal regions of the long bones and infiltration in other organs. The histopathologic hallmark is defined by a mononuclear infiltrate of foamy histiocytes and rare pathognomonic Touton giant cells with extensive fibrosis. This condition is exceptional in children. We report here a case of Erdheim-Chester disease in a 10-year-old girl with retroperitoneal infiltration and bone involvement, for whom the diagnosis was only established after a 3-year course with multiple biopsies. It is also the first pediatric case successfully treated with interferon-alpha suggesting that interferon-alpha can be a safe and efficient first-line therapy for this disease in children.	19755920
2009 Sep	Clin Nucl Med	Intensely hypermetabolic extra-axial brainstem tumor in Erdheim-chester disease	Tan IB, Padhy AK, Thng CH, Osmany S, Magsombol B, Ho YH, Tham CK, Quek R, Tao M, Lim ST	Department of Medical Oncology , National Cancer Centre, Singapore, Singapore	Erdheim-Chester disease is a rare non-Langerhans cell histiocytosis characterized by progressive histiocytic proliferation with multiorgan involvement, typically of the kidney, skin, brain, and lung, and less frequently, the heart and retro-orbital tissue. Fluorine-18 fluorodeoxyglucose positron emission tomography (F-18 FDG PET) plays an important role in the management of this disease. It has been reported that FDG PET imaging allows accurate evaluation of the extent of the disease at baseline, as well as assessment of response to any specific therapy. In this case, a 57-year-old Chinese man presented with functional decline and a urinary tract infection. He had a prior history of xanthogranulomas of bilateral canthal masses. On imaging, he was found to have left hydronephrosis, diffuse urothelial thickening, increased density of the perinephric fat, mural thickening of the descending aorta and soft tissue masses along the posterior wall of the right atrium extending into the region of the interatrial septum and involving the right atrioventricular groove. Histopathology revealed retroperitoneal fibrosis. An IV contrast-enhanced FDG PET scan showed increased activity in a previously unidentified brain stem mass and the shafts of bilateral femora. Varying levels of FDG uptake were seen in the other lesions.	19692824
2009 Aug 11	Leuk Res.	Erdheim-Chester disease: Multisystem involvement and management with interferon-alpha	Suzuki HI, Hosoya N, Miyagawa K, Ota S, Nakashima H, Makita N, Kurokawa M	Department of Hematology and Oncology, Graduate School of Medicine, University of Tokyo, 7-3-1 Hongo, Bunkyo-ku, Tokyo 113-8655, Japan		19679354

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009 Jul 27	Joint Bone Spine	Spumous histiocytic oligoarthritis coexisting with systemic Langerhans' cell histiocytosis: Case report and literature review	Aouba A, Larousserie F, Le Guern V, Martin A, Guillevin L	Department of Internal Medicine, Referal Center for Histiocytosis, Hôpital Cochin, Assistance publique-Hôpitaux de Paris, Université de Paris-5 René-Descartes, 27, rue du Faubourg-Saint-Jacques, 75679 Paris cedex 14, France; Department of Hematology, Hôpital Necker-Enfants-Malades, Assistance publique-Hôpitaux de Paris, Université de Paris-5 René-Descartes, 149, rue de Sèvres, 75473 Paris cedex 15, France.	A 27-year-old man consulted with clinical and radiological features of chronic erosive oligoarthritis of large joints (hips and knee), associated with diffuse lymph-node enlargement and diabetes insipidus. Lymph-node biopsy provided the diagnosis of systemic Langerhans' cell histiocytosis, for which synovial involvement remains a diagnostic challenge. Infectious diseases search and immunological tests were all negative. Skeleton radiographs, hip and cerebral magnetic resonance imaging showed, respectively, erosive arthritis of the hips and stigmates of pituitary-stalk involvement. Hip-synovium biopsy exhibited the main histological features of Erdheim-Chester disease, a non-Langerhans' cell histiocytosis. An extensive literature review found that Langerhans' cell histiocytosis and non-Langerhans' cell histiocytosis (mainly Erdheim-Chester disease) coexistence is rare and synovial involvements in them even more so, these latter presenting mainly as large joint monoarthritis. The absence of typical clinical and radiographic signs of Erdheim-Chester disease led to consideration of the rheumatologic diagnosis of unclassified non-Langerhans' cell histiocytosis (or Erdheim-Chester disease-type) oligoarthritis, associated with multiorgan Langerhans' cell histiocytosis. The differential diagnosis of large joint erosive arthritis should then include both entities, particularly when multiorgan manifestations are present. Non-Langerhans' cell histiocytosis synovial involvements responded poorly to vinblastine and corticosteroids, while Langerhans' cell histiocytosis involvements responded completely but transiently. Both entities regressed under cladribine, with only mild relapses of the non-Langerhans' cell histiocytosis involvements.	19640768
2009 Jul 20	Am J Surg Pathol	Distinctive Pulmonary Histopathology With Increased IgG4-positive Plasma Cells in Patients With Autoimmune Pancreatitis: Report of 6 and 12 Cases With Similar Histopathology	Shrestha B, Sekiguchi H, Colby TV, Graziano P, Aubry MC, Smyrk TC, Feldman AL, Cornell LD, Ryu JH, Chari ST, Dueck AC, Yi ES	Department of Laboratory Medicine and Pathology daggerPulmonary and Critical Care Medicine parallelDepartment of Internal Medicine, Division of Gastroenterology and Hepatology, Mayo Clinic, Rochester, MN double daggerDepartment of Laboratory Medicine and Pathology, Mayo Clinic paragraph signSection of Biostatistics, Mayo Clinic, Scottsdale, AZ section signUnit of Pathology, C. Forlanini Hospital, Roma, Italy	Autoimmune pancreatitis (AP) is one manifestation of a systemic, steroid-responsive disease with elevated serum IgG4 and characteristic histopathology, including increased IgG4-positive (+) plasma cells in the tissue. The histopathology of pulmonary IgG4 disease has not been well established. Six lung biopsies from patients with documented AP were studied, along with 12 additional cases showing similar pulmonary histopathology. For comparison, we examined Erdheim-Chester disease (n=3), pulmonary Sjögren syndrome (n=19), inflammatory myofibroblastic tumor (n=10), various inflammatory and interstitial lung disease (n=61), and nodal or extranodal Rosai-Dorfman disease (RD) in adults (n=8). All cases were stained for IgG4 and scored as 1, 2, and 3 as described in AP according to the following criteria: 0, <5 (per high power field); 1, 5 to10; 2, 11 to 30; and 3, >30. Five lung biopsies from AP patients showed IgG4 score of 3, and 1 had a score of 2. Consistent findings in lung biopsies of AP patients included endothelialitis of pulmonary vessels, active fibrosis, lymphangitic inflammatory infiltrates rich in plasma cells and histiocytes with or without nodule formation, and fibrinous pleuritis. Prominent lymphatic dilatation with histiocytes showing emperipolesis of lymphocytes was also seen. All 12 additional cases showing these histologic features also had the IgG4 score of 2 or 3. Among other conditions, an IgG4 score of 2 or 3 was seen in 6 of 8 RD, 4 of 10 inflammatory myofibroblastic tumors, and 8 of 61 inflammatory and interstitial lung disease, but in none of the rest. In conclusion, distinctive pulmonary histopathology was associated with increased IgG4+ cells in both AP patients and those unknown for AP status. The significance of increased IgG4+ cells in high proportion of RD cases merits further study as does overlap of RD and IgG4 disease.	19623032

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2009 Jul 9	Rheumatol Int.	Erdheim-Chester disease: a case report with pulmonary, kidney involvement and bone lesions.	Mounach A, Nouijai A, Achemlal L, El Maghraoui A, Bezza A.	Military Hospital Mohamed V, Rabat, Morocco, azizamounach@yahoo.fr	We report the case of a 42-year-old woman who was admitted in 2002 for exploration of diffuse bone pain. She had medical history of pulmonary tuberculosis. Her current symptoms had started 9 months earlier and consisted of bone pain, affecting the legs. She had asthenia and weight loss. At admission, physical examination showed bilateral and symmetrical long bone pain, especially the knees and the ankles. Physical exam was normal elsewhere. Laboratory tests showed inflammation, with an erythrocyte sedimentation rate of 90 mm/h and C-reactive protein 8 mg/l. Protein electrophoresis, red and white blood cell count, renal, and liver function tests were normal. Serum calcium, phosphorus, and urinary calcium were normal. Radiographs showed multiple mixed bone lesions with sclerotic and lytic areas of the femora, tibiae, humerus. Chest radiographs and thoracic computed tomography (CT) scan showed pulmonary fibrosis. Biopsy of the tibial area displayed foamy lipid-laden histiocytes, confirming the diagnosis of Erdheim-Chester disease. Patient was treated with prednisolone plus cyclophosphamide. Her clinical condition improved remarkably during 4 years, but she developed acute renal failure leading to death.	19588143
2009 Jul 4	Rheumatol Int.	Erdheim-Chester disease: a pitfall in DXA measurements.	Goerres GW, Gengenbacher MG, Uebelhart D	Institut für Medizinische Radiologie, Buergerspital Solothurn/Spital Grenchen soH, Schoengruenstrasse 42, 4500, Solothurn, Switzerland, ggoerres_so@sec.spital.ktso.ch.	None	19578853
2009 Jun 30	Circulation	Images in cardiovascular medicine. Cardiac involvement in Erdheim-Chester disease: magnetic resonance and computed tomographic scan imaging in a monocentric series of 37 patients.	Haroche J, Cluzel P, Toledano D, Montalescot G, Touitou D, Grenier PA, Piette JC, Amoura Z.	Department of Internal Medicine,Hôpital Pitié-Salpêtrière, 47-83 Boulevard de l'Hôpital, Paris, France. julien.haroche@psl.aphp.fr.	None	19564564
2008 Apr	Tunis Med	Erdheim-Chester disease multivisceral form with favourable outcome	Khanfir A, Moalla H, Boudawarra T, Bahloul A, Mnif J, Abid M, Frikha M.	afefkhanfir@yahoo.fr	None (article in French)	19476146

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2009 Apr	Am J Med Sci.	Erdheim-Chester disease with lung involvement mimicking pulmonary lymphangitic carcinomatosis	Yahng SA, Kang HH, Kim SK, Lee SH, Moon HS, Lee BY, Kim HS, Seo EJ.	Divisions of pulmonology, St. Paul's Hospital, The Catholic University of Korea, Seoul, Republic of Korea.	Erdheim-Chester disease (ECD) is a rare proliferative non-Langerhans cell histiocytosis of multiple organs with unknown etiology. Around 20% of ECD cases are reported to be associated with lung involvement and there are very few cases manifested solely by nonspecific respiratory symptoms. A 50-year-old woman presented with dry cough and dyspnea for 2 weeks. Chest computed tomography (CT) revealed diffuse interlobular septal and fissural thickening with perilymphatic and subpleural nodular opacities, suggesting pulmonary lymphangitic spread of metastatic carcinoma. Bone scintigraphy and positron emission tomography/CT showed multiple skeletal and lymph node involvement. The patient underwent surgical lung biopsy and the pathologic feature was consistent with ECD. We describe this case to emphasize that ECD should be included in the differential diagnosis of cases suspected to have lymphangitic lung carcinomatosis. Moreover, the findings of positron emission tomography/CT scan, which showed hot uptakes in the affected areas, are also described.	19365181
2009 Apr	Chest.	Cardiac tumor and renal involvement in a nonsmoker with centrilobular pulmonary nodules.	Chew HC, Lee CH, Cheah FK, Lim ST, Loo CM.	Department of Respiratory and Critical Care Medicine, Singapore General Hospital, Outram Road, Singapore. chinnjing@pacific.net.sg		19349408
2009 Mar 30	Pathol Res Pract	Clonal status and clinicopathological feature of Erdheim-Chester disease	Gong L, He XL, Li YH, Ren KX, Zhang L, Liu XY, Han XJ, Yao L, Zhu SJ, Lan M, Zhang W.	Department of Pathology, Tangdu Hospital, the Fourth Military Medical University, Shaanxi, Xi'an 710038, China.	Erdheim-Chester disease (ECD) is a rare non-Langerhans form of histiocytosis characterized by xanthomatous tissue infiltration with foamy histiocytes. It is still controversial whether these histiocytic proliferations represent monoclonal neoplastic populations or are part of a polyclonal reactive process. This is a case report of ECD in a 76-year-old Chinese woman. We investigated the clinicopathological features and clonality of the histiocytes using laser microdissection and a clonality assay based on X-chromosomal inactivation mosaicism in female somatic tissues, as well as on the polymorphism of phosphoglycerate kinase (PGK) and androgen receptor (AR). According to our results, the lesion was composed of lipid-laden histiocytes and focal fibrous tissues. The lipid-laden histiocytes were positive for CD68 and CD163, but negative for CD1a and S-100. Electron-microscopic examination showed no Birbeck granules, but the presence of lipid vacuoles. Moreover, the result of the clonality assay demonstrated that these cells formed a polyclonal population. In conclusion, ECD is a rare non-Langerhans' cell histiocytosis. Its nature may be a non-neoplastic lesion; however, additional studies with larger sample sizes are necessary to conclusively prove our hypothesis.	19339122
2009 Mar 19	Eur J Nucl Med Mol Imaging	Symmetric giant xanthogranulomas in Erdheim-Chester disease.	Taguchi T, Sano S, Iwasaki Y, Terada Y.	Kochi Medical School, Kochi University, Kohasu Oko-cho, Nankoku, 783-8505, Japan, tagu@muse.ocn.ne.jp.		19296104

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009 Mar-Apr	Clin Imaging	Erdheim-Chester disease: case report with unique postmortem magnetic resonance imaging, high-resolution radiography, and pathologic correlation.	de Abreu MR, Castro MO, Chung C, Trudell D, Biswal S, Wesselly M, Resnick D.	Department of Radiology, Hospital Mae de Deus, Pedro Chaves Barcelos 1127/401, Porto Alegre RS 90450-010, Brazil. marcelorad@hotmail.com	Erdheim-Chester disease is an infiltrative form of histiocytosis characterized by replacement of normal tissues by lipid-laden histiocytes. The disease typically infiltrates the medullary portion of the diaphysis and metaphysis of long bones, producing a characteristic radiological pattern dominated by bone sclerosis. It usually affects adults of 40 years of age with a clinical spectrum ranging from an asymptomatic focal bone lesion to multisystemic disease. This case report documents unique imaging and pathologic findings of Erdheim-Chester disease using close postmortem pathologic-imaging correlation.	19237062
2009 Apr	Epilepsia, Germany Volume 50, Issue Supplement s4, pages 1–261, April 2009	8th European Congress on Epileptology, Berlin, Germany, 21 – 25 September 2008 - T220 Erdeim-Chester Disease and Epilepsy: Case Report	E. Vitelli, R. Spagliardi, V. Badioni, L. Cucurachi, and M. Riva	Azienda Ospedaliera Della Provincia Di Lodi, Italy	Purpose: Erdheim-Chester disease (ECD) is a rare non-Langerhans histiocytosis with systemic involvement (mainly bone, heart, lung, and kidney). Neurological manifestations are present in 45% of reported patients (Lachenal F et al J Neurol 2006;253:1267–1277) and seizures in 12%. NMR shows different patterns of involvement: infiltrative, meningeal or both. We hereby describe epileptic manifestations and treatment in a case of ECD. Conclusion: Seizures are not surprising in this case of ECD if the diffuse cortical involvement is considered. LEV showed to be effective. We also underline that in the complex treatment of ECD the support of a neurologist is needed, particularly in paucisymptomatic cases, in which the neurological involvement may be subtle or may be the heralding feature.	19356158
2009 Feb	Mov Disord.	Erdheim-Chester disease: a rare clinical presentation as multiple system atrophy.	Chandran V, Pal PK, Moin A, Chickabasaviah YT, Ravishankar S, Panda S.	Pramod Kumar Pal, Department of Neurology, National Institute of Mental Health and Neurosciences, Bangalore, Karnataka, India		19235927
2009, Jan 1	Intern Med.	Cardiac Erdheim-Chester.	Bassou D, El Kharras A, Amezyane T T, En Nouali H, Elbaaj M, Benameur M, Darbi A.	Radiology, Mohammed V Hospital, Rebat, Morocco. d.bassou1966@gmail.com	-	19122364
2008 Dec 30.	Joint Bone Spine.	Erdheim-Chester disease with predominant mesenteric localization: Lack of efficacy of interferon alpha.	Perlat A, Decaux O, Sébillot M, Grosbois B, Desfourneaux V, Meadeb J.	Department of Internal Medicine, Service de médecine Interne, CHU Hôpital Sud, 16 boulevard de Bulgarie, 35200 Rennes, France.	-	19119043

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2009 Jan	Am J Med.	A multiplication problem.	Furlanetto TW, Fischer J, Polanczyk CA, Vasconcelos MV.	Division of Internal Medicine, Hospital de Clínicas de Porto Alegre, Universidade Federal do Rio Grande do Sul, Porto Alegre, RS, Brazil. furlanet@cpovo.net	-	19114169
2008 Nov 22	Med Clin (Barc)	Chemosis due to orbital infiltration in Erdheim- Chester disease	Riancho JA, Gómez-Román J, Hernández JL.	Departamentos de Medicina Interna y Anatomía Patológica. Hospital Universitario Marqués de Valdecilla. Universidad de Cantabria. Santander. Cantabria. España	-	19087836
2008 Sep-Dec	Hell J Nucl Med.	Erdheim-Chester disease: Symmetric uptake in the (99m)Tc-MDP bone scan	Zanglis A, Valsamaki P, Fountos G.	Pammakaristos General Hospital, Nuclear Medicine Department, 43 Iakovaton Street, PC.111 44, Athens, Greece. azanglis@otenet.gr	Erdheim-Chester disease (E-C D) is a rare clinicopathologic entity with nearly pathognomonic radiographic features. About half of the affected exhibit extraskeletal manifestations, including involvement of the hypothalamus-pituitary axis, lung, heart, retroperitoneum, skin, liver, kidneys, spleen and orbit. This disease usually affects individuals in their fifties to their seventies and has a male preponderance. The lesions of E-C D consist of lipid-storing CD68 (+) and CD1a (-) non-Langerhans cell histiocytes, either localized to the bone or involving multiple systems of the body as well. Skeletal involvement is characteristically bilateral and symmetric, exhibiting an osteosclerotic pattern in the metaphysis and diaphysis of the long bones, usually sparing epiphysis. We recently had a 68 years old male patient with E-C D, with a mild and persistent knee pain, who was subjected to a 3-phase technetium-99m methylene diphosphonate ((99m)Tc-MDP) bone-scan and subsequently to gallium-67 citrate ((67)Ga-C) whole body scan. The characteristic symmetric pattern of these scans raised the question of E-CD disease. The patient showed an excellent symptomatic response to high-dose steroids. However, the symptoms recurred after discontinuation of treatment.	19081860
2009 Jan	Brain Pathol.	60-year old woman with extra-axial frontal mass	Arakaki N, Riudavets MA, Cervio A, Ferreira M, Sevlever G.	Institute for Neurological Research, FLENI. Buenos Aires, Argentina	60-year old woman with extra-axial frontal mass.Arakaki N, Riudavets MA, Cervio A, Ferreira M, Sevlever G. We describe a 60 year-old woman presenting with visual loss of her left eye. No lymphadenopathies, fever, or weight loss were detected. Neuroimaging studies revealed an extra-axial mass along the posterior aspect of the left optic nerve. The mass was resected and showed xanthomatous histiocytes that were positive for CD-68, occasionally positive for S-100, and negative for CD-1. The lesion was diagnosed as Erdheim-Chester disease (ECD) affecting the CNS. The patient is under systemic evaluation in order to discover other ECD lesions. Microscopic findings and differential diagnoses are discussed.	19076782

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2008 Nov 25	Rev Neurol (Paris)	Pseudo-tumoral and ischemic encephalic Erdheim-Chester disease	Amezyane T, Abouzahir A, Bassou D, Zoubeir Y, Hammi S, Mahassin F, Ohayon V, Archane MI.	Service de médecine interne B, hôpital militaire d'instruction Mohammed-V, 10000 Hay Ryad, Rabat, Maroc	INTRODUCTION: Erdheim-Chester disease (ECD) is a rare non-langerhans cell histiocytosis of unknown etiology. It is a multi-systematic xanthogranulomatous infiltration with almost constant bone involvement; the neurological manifestations are not specific and occur in 15-20% of cases. METHODS: We report the case of a 59-year-old woman hospitalized for a frontal syndrome and right hemiparesis. RESULTS: Imaging revealed a left caudate nucleus process with recent infarct. Cardiovascular involvement and bilateral osteosclerosis of long bones strongly suggested ECD, confirmed after biopsies of the pericardium and bone. CONCLUSION: Pseudo-tumor encephalic ECD is very rare; the caudate nuclei is an unusual localization; ischemic stroke has been exceptionally described. Prognosis depends largely on the involvement of the central nervous and cardiovascular systems.	19038410
2008 Sep 30	Circulation	Pericarditis Heralding Erdheim-Chester disease.	Vaglio A, Corradi D, Maestri R, Callegari S, Buzio C, Salvarani C.	Department of Clinical Medicine, Nephrology and Health Science, University of Parma, Parma, Italy. augusto.vaglio@virgilio.it		18824648
2008 Sep	Neurol Sci.	Late-onset sporadic ataxia, pontine lesion, and retroperitoneal fibrosis: a case of Erdheim-Chester disease.	Salsano E, Savoiardo M, Nappini S, Maderna E, Pollo B, Chinaglia D, Guerra U, Finocchiaro G, Pareyson D.	Division of Biochemistry and Genetics, IRCCS Foundation, "Carlo Besta" Neurological Institute, Via Celoria 11, 20133, Milan, Italy.	A 60-year-old man with progressive gait ataxia and mild pyramidal signs showed at MRI a pontine lesion with post-contrast enhancement in the left middle cerebellar peduncle. Diagnosis of Erdheim-Chester disease (ECD), a rare non-Langerhans cell histiocytosis, was suggested, further supported by a previously diagnosed retroperitoneal fibrosis. X-ray films demonstrated characteristic bilateral and symmetric osteosclerosis of the long bones of the lower limbs, which at radionuclide studies exhibited a marked increase in technetium-99 uptake. A cerebral 18FDG-PET showed a relevant pontine uptake of the tracer. Re-evaluation of a past retroperitoneal biopsy showed an intense CD68+, CD1a-, and S100-infiltrate of histiocytes with foamy cytoplasm, thus confirming the diagnosis. ECD should be regarded as a rare cause of adult-onset sporadic ataxia, especially when pontine lesions and extraneurological manifestations are present.	18810602
2008	Intern Med.	Erdheim-Chester disease	Vanichaniramol N, Kingpetch K, Buranasupkajorn P, Sunthornyothin S, Snabboon T.	Saraburi Hospital, Saraburi, Thailand.		18797126
2008 Aug 14	N Engl J Med.	Case records of the Massachusetts General Hospital. Case 25-2008. A 43-year-old man with fatigue and lesions in the pituitary and cerebellum.	Mills JA, Gonzalez RG, Jaffe R.	Rheumatology, Allergy, and Immunology Division, Massachusetts General Hospital, Boston, USA.		18703477

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2008 Jul-Aug	Radiographics	Neoplastic and non-neoplastic proliferative disorders of the perirenal space: cross-sectional imaging findings.	Surabhi VR, Menias C, Prasad SR, Patel AH, Nagar A, Dalrymple NC.	Department of Radiology, University of Texas Health Science Center at San Antonio, 7703 Floyd Curl Dr, San Antonio, TX 78229, USA	The perirenal space, located between the anterior and the posterior renal fasciae, is shaped like an inverted cone with an apex that extends into the iliac fossa. Perirenal tumors and pseudotumors primarily originate either from the kidney or as part of a systemic disease process and have characteristic histopathologic features and biologic behavior. The lesions may be classified on the basis of their distribution and imaging features as solitary soft-tissue masses (renal cell carcinoma, lymphangioma, hemangioma, and leiomyoma), rindlike soft-tissue lesions (lymphoma, retroperitoneal fibrosis, and Erdheim-Chester disease), masses containing macroscopic fat (angiomyolipoma, liposarcoma, myelolipoma, and extramedullary hematopoiesis), and multifocal soft-tissue masses (metastases, plasma cell tumors). Because of overlap in imaging findings among these diverse perirenal lesions, a definitive diagnosis in most cases can be established only at histopathologic analysis. However, an imaging pattern-based approach may facilitate the diagnosis and optimal management of perirenal tumors and pseudotumors.	18635626
2008 Jun	Thorax	Progressive dyspnoea, pleural effusions and lytic bone lesions.	Nicholson AG, Anderson E, Saha S, Indrajith M, Conry B, Hughes J.	Department of Histopathology, Royal Brompton Hospital, London, UK.		18511636
2008 May 2	J Thorac Cardiovasc Surg.	Unmasked diabetes insipidus after pericardial drainage and biopsy for pericardial effusion in association with Erdheim-Chester disease.	Augoustides JG, Szeto WY.	Cardiothoracic Section, Anesthesiology and Critical Care, University of Pennsylvania School of Medicine, Philadelphia, 19104-4283, USA. yiandoc@hotmail.com		18603080
2008 Jul 14	J Eur Acad Dermatol Venereol.	Parallel occurrence of Erdheim-Chester disease and eosinophilic granuloma in the same patient.	Kerzl R, Eyerich K, Eberlein B, Hein R, Weichenmeier I, Behrendt H, Clemm C, Fend F, Mempel S, Waldt S, Ring J, Mempel M.	Department of Dermatology and Allergy, Biederstein, Technische Universität München, Germany.		18637864

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2008 May	Rev Port Cardiol	A rare histiocytosis with severe cardiac involvement: Erdheim-Chester disease.	Botelho A, Antunes A, Almeida JC, Abecasis M, de Gouveia RH, Martins AP, Marques AM	Serviço de Cardiologia, Centro Hospitalar de Coimbra, Coimbra, Portugal. a.bota@clix.pt	The term histiocytosis covers various disorders that lead to primary proliferation, infiltration and accumulation of cells of the mononuclear-phagocytic system within the affected tissues. Its pathophysiology is still unclear and the clinical course variable, which explains the lack of specific treatment and the need for a high level of suspicion to arrive at the diagnosis. The authors present the case of a patient with a complex cardiological clinical history, recently referred for surgical treatment of severe mitral insufficiency. Severe thickening of both atrial walls made it impossible to proceed with the intervention. After a complex etiological evaluation, a diagnosis of Erdheim-Chester disease was made. This is a rare, non-Langerhans cell histiocytosis and, to our knowledge, this represents the first case reported in Portugal. The authors also review the literature, particularly of the few cases with cardiac involvement.	18717219
2008 May	Respiration	Erdheim-Chester Disease: Pulmonary Presentation in a Case with Advanced Systemic Involvement.	Protopapadakis C, Antoniou KM, Nicholson AG, Voloudaki A, Tzanakis N, Karantanas A, Siafakas NM	Department of Thoracic Medicine, University of Crete, Heraklion, Greece.	Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytosis usually affecting bone, that may progress to multi-organ involvement, with pulmonary involvement as an indicator of poor prognosis. Herein, we present a 48-year-old man with a 2-year history of progressive exertional dyspnoea, dry cough, malaise and exophthalmos. High-resolution computed tomography showed peripheral interstitial thickening with a lymphangitic distribution throughout both lungs, suspected of representing lymphangitic spread of neoplasia. Transbronchial biopsy specimen and bronchoalveolar lavage were non-diagnostic; thus, a surgical lung biopsy was performed which showed features diagnostic of ECD. Subsequent systematic investigations showed widespread bone involvement, cardiac involvement manifested as left heart failure and renal/perirenal disease. Treatment with pulsed corticosteroids and cyclophosphamide elicited neither clinical nor functional response, with death at 6 months. This case highlights the aggressive nature of ECD when there is pulmonary involvement, as well as problems in diagnosis when there is pulmonary presentation and when systemic disease is asymptomatic.	18460866
2008 May	Ann Nucl Med	Erdheim-Chester disease: a rare syndrome with a characteristic bone scintigraphy pattern.	Spyridonidis TJ, Giannakenas C, Barla P, Apostolopoulos DJ	Department of Nuclear Medicine, Regional University Hospital of Patras, 26500, Rion, Patras, Greece.	Erdheim-Chester disease is a rare noninherited, non-Langerhans' cell histiocytosis, with multiorgan involvement. The skeleton is frequently involved in as many as 70-80% of all cases. In nearly half of the cases, there is an involvement of other organs such as the cardiovascular system, lung, kidneys, brain, and orbits. Extra-skeletal involvement is correlated with increased morbidity and mortality. In recent years, the disease is being described with increasing frequency although fewer than 200 cases have been identified worldwide. Besides its rarity, the disease has a characteristic almost pathognomonic bone scan appearance, which in some cases facilitates diagnosis of the syndrome. Bone scans also contribute to the qualitative assessment of skeletal involvement.	18535884
2008 May	Int J Urol	Retroperitoneal infiltration as the first sign of Erdheim-Chester disease.	**Colin P, Ballereau C, Lambert M, Lemaitre L, Leroy X, Biserte J**	Department of Urology, University Hospital, Lille, France. pierre_colin@msn.com	Case of elderly man with bladder cancer, in whom the first manifestation of Erdheim-Chester disease was retroperitoneal infiltration detected during routine follow-up. The disease was diagnosed on the basis of histology and immunochemistry findings (presence of histiocytes) and of imaging findings (plain radiography, computed tomography, magnetic resonance imaging, and bone scintigraphy). The differential diagnosis with respect to other causes of retroperitoneal infiltration is discussed.	18452465

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2008 Apr	Ann Pathol	Uncommon retroperitoneal and bone lesions: Erdheim-Chester disease.	Mnif H, Makni S, Ayedi L, Trigui W, Bahloul A, Mounir F, Sellami-Boudawara T	Laboratoire d'anatomie et de cytologie pathologiques, CHU Habib-Bourguiba, 3029 Sfax, Tunisie.	We report a case of Erdheim-Chester disease, revealed by a polyuropolydipsic syndrome. During the patient's work-up, osteocondensing lesions were found; the biopsy of these lesions showed an infiltration by spumous cells of histiocytic lineage, CD68+, CD1a-, associated with a lymphoid infiltrate within an extensive fibrosis. Lung and retroperitoneal lesions were discovered. The surgical resection of the involved ureter was required. Histological examination of the resected specimen showed the same pattern of histiocytic infiltration. Our case report underlines the variety of lesions associated with Erdheim-Chester disease and the importance of a complete exploration.	18675171
2008 Feb	Endocr J	Erdheim-Chester Disease: Report of a Case with PCR-based Analysis of the Expression of Osteopontin and Survivin in Xanthogranulomas Following Glucocorticoid Treatment.	**Taguchi T, Iwasaki Y,** Asaba K, Yoshida T, Takao T, Ikeno F, Nakajima H, Kodama H, Hashimoto K	Departments of Endocrinology, Metabolism, and Nephrology, Kochi Medical School, Kochi University.	Case of ECD presenting diabetes insipidus and multiple xanthogranulomas received glucocorticoid treatment over a year with improvement seen. Results suggest that the expression level of osteopontin could be a marker of the disease activity of ECD.	18270430
2008	Skinmed	Erdheim-chester disease with cutaneous features in an Indian patient.	Garg T, **Chander R**, Gupta T, Mendiratta V, Jain M	From the Department of Dermatology, Venereology and Leprosy Lady Hardinge Medical College, New Delhi, India.	Case of 60-year-old Indian woman presented with multiple asymptomatic, firm swellings over the face that had been progressively increasing for the past 3.5 years. She complained of dry cough and dyspnea of 2 years' duration, which was diagnosed as interstitial lung disease (ILD) based on chest radiography and high-resolution computed tomography. Cutaneous examination revealed multiple (5) firm, yellowish to skin-colored well-defined nodules with irregular margins ranging in size from 1x 1 cm to 4x8 cm present over the left periorbital region and right jawline, with overlying telangiectasias on the skin. 40 mg oral prednisolone daily was started. Surgical debulking of her skin lesions was planned, but the patient refused due to her worsening ILD.	18327007
2008 Jan	Virchows Arch	Systemic Erdheim-Chester disease.	Dickson BC, Pethe V, Chung CT, Howarth DJ, **Bilbao JM**, Fornasier VL, Streutker CJ, Sugar LM, **Bapat B**	Department of Pathology and Laboratory Medicine, Mount Sinai Hospital, Toronto, ON, Canada.	Clinical histories, pathologic findings, and an analysis of clonality using the HUMARA assay in two patients diagnosed with Erdheim-Chester disease. One case has previously been documented in the literature. Histologically, both cases demonstrated sheets of foamy xanthomatous histiocytes with widespread infiltration of the viscera. We demonstrate the histiocytes to express CD163, thereby further supporting a monocyte/macrophage basis. Moreover, in confirming clonality, our observations lend additional evidence to the view that Erdheim-Chester disease represents a neoplastic process.	18188596
2008 Jan	Nat Clin Pract Rheumatol	A case of Erdheim-Chester disease initially mistaken for Ormond's disease.	Loddenkemper K, Hoyer B, Loddenkemper C, Hermann KG, Rogalla P, Förster G, Buttgereit F, Hiepe F, Burmester GR	Department of Rheumatology and Clinical Immunology, Charité University Medicine, Berlin, Germany. konstanze.loddenkemper@charite.de	A 54-year-old man presented with fever, abdominal pain, anemia, elevated C-reactive protein level and decreased renal function. Idiopathic retroperitoneal fibrosis (Ormond's disease) had been diagnosed in the past, leading to surgical ureterolysis. Further testing led to a diagnosis of Erdheim-Chester disease with retroperitoneal fibrosis and bone sclerosis. Treatment with glucocorticoids failed. The patient's symptoms improved significantly after initiation of interferon-alpha therapy.	18172449

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2007 Nov	Clin Nucl Med	FDG PET/CT for Biopsy Guidance in Erdheim-Chester Disease.	E Lin	From the Department of Radiology, Virginia Mason Medical Center, Seattle, WA.	A 60-year-old man with a history of non-Hodgkin lymphoma underwent FDG PET/CT which demonstrated a focal area of uptake in the left posterior perirenal space, and uptake in both hips. The focal area of uptake in the left perirenal space was biopsied, which demonstrated Erdheim-Chester disease. The PET/CT was important in identifying an area for biopsy and demonstrating bone involvement.	18075421
2007 July	Presse Med	[Erdheim-Chester disease.]	Haroche J, Amoura Z, Wechsler B, Veyssier-Belot C, Charlotte F, Piette JC	Service de médecine interne, Hôpital Pitié-Salpêtrière, Paris (75).	Erdheim-Chester disease classically thought to be rare, but diagnosed more frequently nowadays (250 published cases). Two signs highly evocative of this diagnosis are nearly constant tracer uptake by the long bones on (99)Tc bone scintigraphy and a "hairy kidney" appearance on abdominal CT scan. A more "elegant" diagnostic method is ultrasound-guided biopsy of the perirenal infiltration. Cardiovascular involvement, which affects the aorta ("coated aorta") as well as all the cardiac layers, leads to one third of the deaths related to this disease. Central nervous system infiltration (especially cerebellar) is severe and difficult to treat. The prognosis is extremely variable and is often worse when there is a cardiovascular and/or central nervous system involvement. The treatment, decided upon on a case-by-case basis at a specialist center, often begins with interferon alpha.	17618076
2007 Jun	Rev Med Interne	[Usefulness of combined positron emission tomography and computed tomography imaging in Erdheim-Chester disease.]	Girszyn N, Arnaud L, Villain D, Kahn JE, Piette AM, Bletry O	Service de médecine interne, hôpital Foch, 40, rue Worth, 92151 Suresnes cedex, France.	Use of combined fluorodeoxyglucose positron emission tomography and computed tomography (18F-FDG PET-CT) in this disease is reported. EXEGESIS: Three men, aged from 55 to 74 years with confirmed Erdheim-Chester disease were included. 18F-FDG PET-CT allowed to detect visceral and vascular involvement of the disease which were overlooked with CT-scan or magnetic resonance imaging: left common carotid and ilio-femoral artery in one patient, coronary, femoral and tibia in the second, aortic, common carotid, femoral and mandibula in the remaining patient. Also, sequential 18F-FDG PET-CT was useful to appreciate treatment efficiency (decrease hyperfixation) and decide treatment modification (interferon alpha). CONCLUSION: 18F-FDG PET-CT combined imaging allows to assess the extent of involvement in Erdheim-Chester disease. 18F-FDG PET-CT may be also a useful tool in the management of Erdheim-Chester disease.	17629593
2007 Oct	Australas Radiol	Erdheim-Chester disease: a rare cause of acute renal failure.	O'Rourke R, Wong DC, Fleming S, Walker D	Radiology Department, The Wesley Hospital, Brisbane, Queensland, Australia.	Report one case that presented with an encased aorta and renal arteries leading to acute renal failure. The diagnosis of ECD was delayed until a biopsy of the retroperitoneal infiltrate was performed. Further imaging with fluorine 18 deoxyglucose positron emission tomography, bone scintigraphy, plain films of the long bones and CT of the chest, abdomen and pelvis were performed to assess the extent of the patient's systemic disease involvement. To our knowledge, this is the first reported case of ECD presenting with acute renal failure secondary to bilateral occlusion of the renal arteries.	17875157
2007 Oct	Arq Bras Oftalmol	[Intraocular involvement in Erdheim-Chester disease - first report in the literature: case report.]	Biccas Neto L, Zanetti F	Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil.	This is the first report of intraocular involvement in this disease. MPSG, a 46 y.o. woman, presented with proptosis of the OD. She referred ulcerated lesions on the hard palate, symmetrical and bilateral osteosclerosis of the fibulae and tibiae and a nodule in the right breast (biopsy: xantomatous histiocytic infiltrate CD68+, S-100 and CD1a negative on immunohistochemistry compatible with ECD). MRI studies demonstrated an extraconal tumor in the juxta-bulbar temporal portion of the right orbit close to the lacrimal gland and hyperintense on T1. This pioneer report depicts in vivo characteristics of histiocytic granulomas in ECD. Caution should be taken with patients with ECD as potentially blinding intraocular complications may arise.	18157316

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2007 May	Skeletal Radiol	An unusual case of Erdheim-Chester disease with features of Langerhans cell histiocytosis.	Furmanczyk PS, Bruckner JD, Gillespy T, Rubin BP	Department of Pathology, University of Washington Medical Center, 1959 NE Pacific, Room BB220, P.O. Box 356100, Seattle, WA, 98195-6100, USA, pfurman@u.washington.edu.	We report on a case of ECD with some features suggestive of LCH. Radiographs demonstrated a large lytic lesion in the left femur, with multiple lesions of sclerosis involving both distal femurs and tibias. Both the lytic lesion and a sclerotic lesion were biopsied and demonstrated distinctive histologic features characteristic of ECD in the tibia and features of LCH in the femur. The clinical/radiologic and pathologic features that distinguish ECD and LCH as distinct entities are reviewed, and the underlying biological connection between them is discussed.	17492445
2007 Jun	Coll Antropol	Erdheim-Chester disease and concomitant tuberculosis successfully treated with chemotherapy and long-term steroids.	Badzek S, Misir-Krpan A, Krajina Z, Radman I, Stern-Padovan R, Dotlić S	Department of Oncology, University Hospital Center "Zagreb", Zagreb, Croatia. sbadzek@kbc-zagreb.hr	According to published material and our experience, cytotoxic chemotherapy and long-term steroids have therapeutic benefit. Although this approach can probably be accepted as standard of care management, novel therapeutic modalities should be explored, and pathogenesis and disorder classification should be cleared out as well. The case of ECD affecting skeletal system and lungs and concomitant laryngeal tuberculosis successfully treated with chemotherapy and long-term steroid therapy is presented.	17847948
2007 Jun	Hong Kong Med J	Orbital involvement in Erdheim-Chester disease.	Lau WW, Chan E, Chan CW	Department of Ophthalmology, Queen Mary Hospital, Pokfulam Road, Hong Kong.	A 45-year-old woman presenting with unilateral proptosis and periorbital xanthelasma. Histopathological examination revealed a xanthogranulomatous lesion expressing CD68, but negative for S100 protein, CD1a, CD3, or CD20. Systemic involvement was evident on bone scanning, and involvement of the thorax and abdominal aorta was seen on computed tomography. Despite treatment with systemic steroids, immunosuppressants, chemotherapy and interferon, progressive deterioration occurred. Our patient's clinical course was consistent with reports in the literature. Unfortunately, our patient developed neutropenic fever and died from septicaemic shock.	17548915
2007 June	Archives of Dermatology	Imatinib as a Treatment Option for Systemic Non-Langerhans Cell Histiocytoses	Jochen Utikal, MD; Selma Ugurel, MD; Hjalmar Kurzen, MD; Philipp Erben, MD; Andreas Reiter, MD; Andreas Hochhaus, MD; Thomas Nebe, MD; Ralf Hildenbrand, MD; Uwe Haberkorn, MD; Sergij Goerdt, MD; Dirk Schadendorf, MD	Jochen Utikal, MD, Massachusetts General Hospital Cancer Center and Harvard Stem Cell Institute, 185 Cambridge St, Boston, MA 02114 (jutikal@mgh.harvard.edu).	Herein, we report the case of a 41-year-old man with Rosai-Dorfman disease, a form of systemic non-Langerhans cell histiocytoses, with histiocytic infiltrations in the skin, bone marrow, liver, and spleen. Histiocytes were positive for the imatinib target proteins platelet-derived growth factor receptor β and KIT. The disease completely responded to treatment with 400 to 600 mg daily of imatinib for more than 7 months.	

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2007 Jun	Int J Clin Oncol	Radiotherapy for Erdheim-Chester disease.	Matsui K, Nagata Y, Hiraoka M	Department of Radiology, Maizuru Municipal Hospital, Kyoto, Japan, k-matsui@hospital.toyooka.hyogo.jp.	A 42-year-old woman suffered from pain in both legs, and fever. She was diagnosed with Erdheim-Chester disease (ECD), based upon bone biopsy. Initially, she received steroid therapy, which led to temporary improvement. However, bone pain in the right femur was so progressive that, as a trial course of radiation therapy, she was given a total dose of 18 Gy in ten fractions to the right distal femur. She showed a gradual response, and the local pain became controllable.	17566851
2007 Apr	J Clin Endocrinol Metab	Bilateral Adrenal Infiltration in Erdheim-Chester Disease. Report of Seven Cases and Literature Review.	Haroche J, Amoura Z, Touraine P, Seilhean D, Graef C, Birmelé B, Wechsler B, Cluzel P, Grenier PA, Piette JC	From Service de Médecine Interne, Service d'Endocrinologie et Médecine de la Reproduction, Service de Neuropathologie, and Service de Radiologie Hôpital Pitié-Salpêtrière, 47-83 Bld de l'Hôpital, 75013 Paris, France; Service de Néphrologie - Immunologie Clinique, Hôpital Bretonneau, 2 Boulevard Tonnelé, 37044 Tours, France.	22 patients with ECD undergoing systematic computed tomography (CT) scan to search for signs of adrenal enlargement. Results: Seven of the 22 (31.8%) patients with ECD displayed adrenal infiltration on CT scan. In one case, autopsy confirmed that the adrenal enlargement was due to foamy histiocyte infiltration in the adrenal glands. Adrenal involvement was reported in only 15 of the 240 ECD cases published up to May 2006. This frequency is significantly lower than that in our series (p = 0.0008; Fisher's exact test). Conclusions: Physicians should be aware of ECD as a possible cause of morphological changes in adrenal size and infiltration.	17405844
2007 May	Brain Dev	Erdheim Chester disease: cerebral involvement in childhood.	Kumandaş S, Kurtsoy A, Canöz O, Patiroğlu T, Yikilmaz A, Per H	Department of Pediatric Neurology, Faculty of Medicine, Erciyes University, 38039 Kayseri, Turkey. skumandas@hotmail.com	We reported the case of a 10-year-old boy who presented headache, paraparesis and with diabetes inspidus for 6 years. As far as we know, the case presented here is the first published report of intracranial involvement and unilateral bone sclerosis with ECD in childhood.	17014978
2007 May	Oral Surg Oral Med Oral Pathol Oral Radiol Endod	Oral radiographic and clinico-pathologic presentation of Erdheim-Chester disease: a case report.	Dinkar AD, Spadigam A, Sahai S	Oral Medicine, Diagnosis, and Radiology Department, Goa Dental College and Hospital (Government of Goa), Bambolim, Goa, India.	A 69-year-old woman with unexplained fever and weakness was referred for evaluation of a solitary mandibular swelling adjacent to a severely resorbed edentulous mandibular ridge. The patient had coexisting craniofacial-skeletal lesions and diabetes insipidus. Histological and immunohistochemical staining of sections from mandibular lesions confirmed the rare diagnosis of Erdheim-Chester disease. The absence of cardiac, pulmonary, renal, and major neurological manifestations was suggestive of a diagnosis at an early stage of the disease. Early diagnosis has been rare with less than 100 reported cases. A review of the literature revealed only 2 cases that report detailed maxillomandibular radiographic findings. A seemingly benign clinical presentation of a potentially grave disease that presents with an osteolytic-sclerotic oral radiographic picture is reported.	17317237

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2007 Apr	Acta Chir Orthop Traumatol Cech	[Warfarin-induced hemorrhagic pseudocyst in the pelvic of a woman with an inherited disorder of blood coagulation, complicated by pelvic bone pseudoxanthoma mimicking erdheim-chester disease.]	Kinkor Z, Koudela K, Koudela K, Havlícek F, Koudelová J	Bioptická laborator, s.r.o., Plzen	A 50-year-old woman with developmental dysplasia of the hip underwent total hip arhtroplasty, and subsequently developed recurrent venous thrombophilia of the lower extremities. Hematological examination revealed an inherited disorder of blood coagulation (homozygous mutation of the 5,10-methylenetetrahydrofolate reductase gene) and therefore longterm Warfarin anticoagulation therapy was started. A year later she was diagnosed with a large pelvic posthemorrhagic pseudocyst (hematoma) located below the musculus iliacus and adhering to bone in the region of posterior acetabulum. The condition was complicated by usuration and focal osteolysis of the adjacent pelvic bone. Histological examination of the hematoma showed characteristics of an unusual pseudoxanthoma mimicking Erdheim-Chester disease. The differential diagnosis of histological findings is discussed and recent relevant literature is reviewed. Key words: warfarin-induced hematoma, posthemorrhagic pseudocyst, musculus iliacus, pelvis, anticoagulation therapy, pseudoxanthoma of the bone, Erdheim-Chester disease.	17493413
2007 Mar	J Neurooncol	Cerebral Erdheim-Chester disease: first report of child with slowly progressive cerebellar syndrome.	Ozdemir MA, Coşkun A, Torun YA, Canoz O, Kurtsoy A, Patıroğlu T	Department of Pediatric Hematology, Erciyes University Medical School, Talas C, Kayseri, 38039, Turkey, makifo@erciyes.edu.tr.	Age at diagnosis ranges from 7 to 84 years (mean age, 53 years) with a female-to-male ratio of 3:1. Pediatric cases are extremely rare based on a search of the English-language literature, and only three cases have been reported; they were in a 7-, 10- and a 14-year-old. We described a 10-year-old boy with ECD who showed six years clinical course of slowly progressive cerebellar symptoms. To our knowledge, this may be the first case of a slowly progressive cerebellar syndrome associated with ECD in a child.	17361336
2007 Feb	Am J Gastroenterol	Biliary manifestation of Erdheim-Chester disease mimicking Klatskin's carcinoma.	Gundling F, Nerlich A, Heitland WU, Schepp W	Second Department of Medicine, Bogenhausen Academic Teaching Hospital, Technical University of Munich, Munich, Germany.	We report a patient with elevated serum levels of liver enzymes due to intra- and extrahepatic bile duct stenoses. The patient's past medical history was remarkable for ECD, since 1 yr before he had undergone surgery for a pituitary lesion in our neurosurgical department revealing the typical histological and immunohistochemical criteria of ECD. Because no biliary manifestation of ECD had been described so far in the literature, surgery of suspected bile duct carcinoma was performed unraveling an unresectable tumor of the hilar region. Surprisingly, histologic examination of intraoperative biopsy specimens failed to demonstrate malignancy but rather revealed another xanthogranulomatous lesion embedded in extended periductal fibrosis as is typically described in extrahepatic parenchymal organ manifestation of ECD. Other possible reasons for cholestatic liver disease were excluded. Secondary cholestasis was overcome by endoscopic dilatation and biliary stenting with stents being exchanged every 3 months. During follow-up for 7 yr we have observed only a slight increase of the hilar stenosis so far. This is the first report describing biliary manifestation of ECD. Even though ECD is a rare cause of cholestasis, it should be considered in patients with this disorder in the setting of multiorgan manifestation.	17037989
2007 Feb	Am J Surg Pathol	Clonal cytogenetic abnormalities in Erdheim-Chester disease.	Vencio EF, Jenkins RB, Schiller JL, Huynh TV, Wenger DD, Inwards CY, Oliveira AM	Division of Anatomic Pathology, Mayo Clinic, Rochester, MN 55905, USA.	We report for the first time the cytogenetic findings of a case of ECD diagnosed at Mayo Clinic Rochester. The tumor occurred in the right tibia of a 35-year-old man and showed the balanced chromosomal translocation t(12;15;20)(q11;q24;p13.3), among other numeric chromosomal abnormalities. The lesion was positive for CD68 and negative for CD1a and S100. These findings support the idea that some cases of ECD are clonal neoplastic disorders of putative histiocytic differentiation. However, additional studies are warranted to confirm whether the chromosomal abnormalities found in this case represent recurrent cytogenetic events.	17255779

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2007 Feb	Singapore Med J	Erdheim-Chester disease: a rare cause of interstitial lung disease.	Kong PM, Pinheiro L, Kaw G, Sittampalam K, Teo CH	Department of Respiratory Medicine, Tan Tock Seng Hospital, 11 Jalan Tan Tock Seng, Singapore 308433. po_marn_kong@ttsh.com.sg	ECD should be considered in the differential diagnosis of interstitial lung disease. We describe a 39-year-old woman who presented with dry cough, malaise and progressive dyspnoea. She was diagnosed to have late stage interstitial lung disease due to Erdheim-Chester disease.	17304381
2007 Jan	Nihon Kokyuki Gakkai Zasshi	[A case of Erdheim-Chester disease effectively treated by cyclophosphamide and prednisolone]	Yano S, Kobayashi K, Kato K, Tokuda Y, Ikeda T, Takeyama H	Department of Pulmonary Medicine, National Hospital Organization Matsue National Hospital.	We report a 55-year-old man with ECD who complained of severe dyspnea despite home oxygen therapy with noninvasive positive pressure ventilation. Continuous PGI2 administration was not very effective, but administration of cyclophosphamide and prednisolone induced rapid improvement of respiratory failure and the effect for six months on arterial blood gas analysis and stability of the disease state persisted.	17313026
2007 Jan	Nucl Med Commun	Radiopharmaceutical diagnosis of Erdheim-Chester's disease.	Palotás A, Bogáts G, Lázár M, Papós M, Matin K, Pávics L	Division of Cardiac Center for Cardiology Department of Psychiatry, Asklepios-Med Bt., H-6722 Szeged, Kossuth Lajos sgt. 23, Hungary. palotas@nepsy.szote.u-szeged.hu	We have previously suggested diagnostic methods using radioisotopes to evaluate this disseminating disease, but they are neither specific nor selective in this regard. The present hypothesis-driven paper reviewing our case proposes novel approaches involving nuclear medicine and utilizing radiopharmaceuticals to identify this potentially fatal multi-system disease.	17159551
2007 Jan	Rheumatol Int	Treatment of skeletal Erdheim-Chester disease with zoledronic acid: case report and proposed mechanisms of action.	Srikulmontree T, Massey HD, Roberts WN	Rheumatology Section, Hunter Holmes McGuire Medical Center, 1201 Broad Rock Blvd, 111M, Richmond, VA, 23249, USA.	Here we report a case of biopsy-proven skeletal ECD, who received treatment with zoledronic acid, an aminobisphosphonate, with remarkable clinical improvement. We also discuss possible mechanisms of action of bisphosphonates in this disorder, especially their roles in inhibition of inflammatory cytokines and macrophage infiltration.	16932956
2006 Dec	Arthritis Rheum	Immunohistochemical evidence of a cytokine and chemokine network in three patients with Erdheim-Chester disease: implications for pathogenesis.	Stoppacciaro A, Ferrarini M, Salmaggi C, Colarossi C, Praderio L, Tresoldi M, Beretta AA, Sabbadini MG	University of Rome La Sapienza, Rome, Italy.	The purpose of this study was to assess cell proliferation and expression of cytokines, chemokines, and chemokine receptors that may potentially be important in histiocyte accumulation in ECD lesions. Biopsies were performed on 3 patients with ECD. Our data indicate that, similar to LCH, ECD lesions are characterized by a complex cytokine and chemokine network, which may orchestrate histiocyte activation and accumulation through an autocrine loop and contribute to the pathogenesis of the disease.	17133532
2006 Dec	J Cardiovasc Pharmacol Ther	An isotope-diagnostic approach to Erdheim-Chester's disease of the heart.	Palotás A, Bogáts G, Lázár M, Papós M, Matin K, Pávics L	Division of Cardiac Surgery, Center for Cardiology, Albert Szent-Györgyi Medical and Pharmaceutical Center, Faculty of Medicine, University of Szeged, Szeged, Hungary. palotas@nepsy.szote.u-szeged.hu	We present several specific isotope-diagnostic techniques of a case to support the identification of this rare multisystem infiltrative disease.	17220475

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2006 Oct	Arthritis Rheum	Variability in the efficacy of interferon-alpha in Erdheim-Chester disease by patient and site of involvement: results in eight patients.	Haroche J, Amoura Z, Trad SG, Wechsler B, Cluzel P, Grenier PA, Piette JC	Hôpital Pitié-Salpêtrière, Paris, France. julien.haroche@psl.ap-hop-paris.fr	We treated 8 patients with multisystemic ECD with subcutaneous interferon-alpha (IFNalpha) at a dosage of 3-9 x 10(6) units 3 times weekly, for a median duration of 23 months (range 1-46 months). RESULTS: Treatment was generally well tolerated, and side effects remained limited to fever following injections. Treatment was discontinued in 1 patient, because of severe depression. During treatment, some manifestations of ECD disappeared (i.e., xanthelasma, exophthalmos, papilledema, and intracranial hypertension). The efficacy of IFNalpha on cardiovascular ECD was variable, however. Treatment resulted in partial regression of "coated aorta" in some cases and clear failure in others; 2 patients died. The level of C-reactive protein diminished sharply in 5 patients. CONCLUSION: IFNalpha might be a valuable first-line therapy for prolonged treatment of ECD. However, the efficacy of IFNalpha varies among patients and according to the sites of disease involvement, and symptoms may fail to respond to treatment, especially in patients with severe multisystemic forms of ECD with central nervous system and cardiovascular involvement.	17009306
2006 Oct	J Neurol	Neurological manifestations and neuroradiological presentation of Erdheim-Chester disease: report of 6 cases and systematic review of the literature.	Lachenal F, Cotton F, Desmurs-Clavel H, Haroche J, Taillia H, Magy N, Hamidou M, Salvatierra J, Piette JC, Vital-Durand D, Rousset H	Department of Internal Medicine, Centre Hospitalier Lyon Sud, 69495, Pierre-Bénite, Cedex, France. flo.lachenal@free.fr	We report 6 cases of ECD with neurological involvement and neuroradiological abnormalities on brain MRI. A literature review revealed 60 other cases of ECD with neurological involvement. We therefore analyzed 66 ECD patients with neurological involvement. Cerebellar and pyramidal syndromes were the most frequent clinical manifestations (41% and 45% of cases), but seizures, headaches, neuropsychiatric or cognitive troubles, sensory disturbances, cranial nerve paralysis or asymptomatic lesions were also reported. Neurological manifestations were always associated with other organ involvement, especially of bones (at least 86%) and diabetes insipidus (47%). Neurological involvement was responsible for severe functional handicaps in almost all patients and was responsible for the death of 6 of the 66 patients (9%). Neuroradiological findings could be separated into three patterns: the infiltrative pattern (44%), with widespread lesions, nodules or intracerebral masses, the meningeal pattern (37%), with either thickening of the dura mater or meningioma-like tumors, and the composite pattern (19%), with both infiltrative and meningeal lesions.	17063320
2006	Neurol Neurochir Pol	Neurologic presentation of Erdheim-Chester disease.	Brodkin CL, Wszolek ZK	Department of Neurology, Mayo Clinic, 4500 San Pablo Road, Jacksonville, FL 32224, USA.	We present 2 cases and reviewed 108 patients reported in the literature who had neurologic manifestations of Erdheim-Chester disease. After eye involvement or diabetes insipidus, cerebellar symptoms were most frequently encountered, followed by tumor, headaches, cord compression, mental status change, seizures, and change in libido. A wide range of neurological symptoms can be seen in ECD. Therefore we hope the review brings more awareness about this disorder.	17103353
2006	Radiologia	[Radiologic diagnosis of Erdheim-Chester disease. A case report]	Gil Marculeta R, Domínguez Echávarri PD, Cano Rafart D, Larrache Latasa J	Servicio de Radiología, Clínica Universitaria de Navarra, Pamplona, España. rgil@unav.es	Erdheim-Chester disease is a rare disorder, belonging to the group of histiocytoses, in which diffuse infiltration of histiocytes affects various organs and systems. Bone involvement in Erdheim-Chester disease manifests as generalized sclerosis of the bone marrow and cortex of the long bones, and this peculiar radiologic characteristic differentiates it from other histiocytoses. Diagnostic suspicion of the disease derives from the pulmonary and bone radiologic findings as well as from the clinical findings. Histological study reveals histiocyte infiltration affecting the soft tissues, musculoskeletal system, and central nervous system. The definitive diagnosis is reached by immunohistochemistry. Like other histiocytoses, such as Langerhans cell histiocytosis, immunohistochemical techniques reveal lipid-laden histiocytes; however, unlike the other types, Erdheim-Chester histiocytes stain negatively for S 100 protein and do not contain Birbeck granules.	17168244

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2006 Sep	Radiother Oncol	Palliative treatment of Erdheim-Chester disease with radiotherapy: a Rare Cancer Network study.	Miller RC, Villà S, Kamer S, Pasquier D, Poortmans P, Micke O, Call TG	Department of Radiation Oncology, Mayo Clinic, Rochester, MN 55905, USA.	A retrospective study of the use of palliative external beam radiotherapy (EBRT) in nine patients with Erdheim-Chester disease was conducted through the Rare Cancer Network. Patients received EBRT for bone pain, brain infiltration, or retro-orbital involvement. EBRT typically provided short-term palliation, with later recurrence of symptoms in most cases.	16959346
2006 Jul	Med Klin (Munich)	[Erdheim-chester disease: a rare cause of interstitial lung disease]	Krüger S, Krop C, Wibmer T, Pauls S, Mottaghy FM, Schumann C, Hombach V	Innere Medizin II, Universitätsklinikum Ulm, Ulm. s.krueger@uniklinik-ulm.de	A 58-year-old man presented with fatigue, diffuse pain of the lower extremities, dyspnea, and a dry cough. CT demonstrated pulmonary fibrosis, periaortic fibrosis of the thoracic aorta, and retroperitoneal fibrosis. The diagnosis of Erdheim-Chester disease was confirmed by minimally invasive lung biopsy. Steroid therapy was not tolerated. Following a stable interval of 18 months there was a disease progression, which could be stabilized after the initiation of cyclophosphamide therapy. CONCLUSION: In patients with extensive pulmonary fibrosis and coincidence of other organ manifestations such as periaortic or retroperitoneal fibrosis and particularly in case of symmetrical osteosclerotic bone lesions, Erdheim-Chester disease should be considered. Immunosuppressive therapy can lead to a stabilization or even improvement of the disease.	16850173
2006 Apr	Laryngoscope	Subglottic stenosis in Erdheim-Chester disease: a previously unrecognized site of involvement.	Freed GL, Sinacori JT	Department of Otolaryngology-Head and Neck Surgery, Eastern Virginia Medical School, Norfolk, Virginia 23507, USA.	We describe a case of laryngeal stenosis secondary to an etiology not previously described. A patient with Erdheim-Chester disease presented with airway obstruction and was found to have subglottic stenosis. Biopsy results confirmed Erdheim-Chester nodules as the cause of the obstruction. This case illustrates the need for biopsy to rule out malignancy and less common etiologies of subglottic stenosis.	16585877
2006 Feb	Radiology	Bone involvement in Erdheim-Chester disease: imaging findings including periostitis and partial epiphyseal involvement.	Dion E, Graef C, Miquel A, Haroche J, Wechsler B, Amoura Z, Zeitoun D, Grenier PA, Piette JC, Laredo JD	Department of Radiology, La Pitié Salpêtrière Hospital, 47-83 Boulevard de l'Hôpital, 75651 Paris Cedex 13, France. elisabeth.dion@psi.ap-hop-paris.fr	PURPOSE: To retrospectively review the bone findings at radiography, scintigraphy, computed tomography (CT), and magnetic resonance (MR) imaging in 11 patients with immunohistochemical and histologic proof of Erdheim-Chester disease. CONCLUSION: This series provides a detailed description of bone involvement in Erdheim-Chester disease. Periostitis and partial epiphyseal involvement of the long bones are also features of this disease. (c) RSNA, 2005.	16371583
2006	Heart Surg Forum	Erdheim-Chester's disease of the heart: a diagnostic conundrum and collision with the same mass in the orbit.	Bogáts G, Piros G, Tiszlavicz L, Iványi B, Sasi V, Csepregil L, Simon J, Babik B, Csillik A, Kardos L, Palkó A, Matin K, Hanzély Z, Korányi K, Nyáry I, Végh M, Kolozsvári L, Kahán Z, Bajcsay A, Tóth A, Balázs G, Simor T, Pávics L, Palotás A	Division of Cardiac Surgery, Center for Cardiology, Faculty of Medicine, University of Szeged, Szeged, Hungary.	Erdheim-Chester's disease is a rare multisystem xanthogranulomatosis, afflicting the skeletal system with the occasional involvement of soft tissues. We delineate an unusual case of a cardiac variant of Erdheim-Chester's disease presenting with pericardial effusion and as a collision with a synchronous orbital manifestation. We describe our diagnostic pathway and propose a novel treatment option involving nonsteroidal anti-inflammatory drugs. The role of cyclo-oxygenase in the disease process and inhibition thereof by NSAIDs is hypothesized and discussed.	16403713

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2006	Ophthal Plast Reconstr Surg	Association between Erdheim-Chester disease, Hashimoto thyroiditis, and familial thrombocytopenia.	Cruz AA, de Alencar VM, Falcão MF, Elias J, Chahud F	Department of Ophthalmology, Otorhinolaryngology, and Head and Neck Surgery, School of Medicine of Ribeirão Preto, University of São Paulo, Brazil. aavecruz@fmrp.usp.br	A 28-year-old woman presented with progressive proptosis of the left eye. She had a history of familial thrombocytopenia and Hashimoto thyroiditis. A review of the literature indicated that the association between non-Langerhans histocytoses and immunologic dysfunctions is not uncommon. We hypothesize that Erdheim-Chester disease may be linked to an abnormal interaction between T-lymphocytes and macrophages similarly to the macrophage activation syndromes.	16418672
2005 Dec	Recenti Prog Med	[Erdheim-Chester disease: normal skeletal radiography in a patient with extensive bone involvement]	Gabrielli GB, Stanzial AM, Moretti L, Volpe A, Corrocher R	Dipartimento di Medicina Clinica e Sperimentale, Università di Verona.	The patient we describe suffered of serious clinical symptoms in the lower limbs, but the direct radiography of the legs did not show any abnormality; this finding seems very remarkable and, to our knowledge, has not been reported previously in the literature. Therefore we discuss the role of the imaging procedures in the diagnosis of Erdheim-Chester disease. Differently from other authors, we did not obtain any clinical improvement in our patient by steroid treatment alone, that is generally considered the first therapeutic option for Erdheim-Chester disease with only skeletal involvement.	16496745
2005 Nov	Blood	Successful treatment of Erdheim-Chester disease, a non-Langerhans-cell histiocytosis, with interferon-alpha.	Braiteh F, Boxrud C, Esmaeli B, Kurzrock R	Phase I Program, Division of Cancer Medicine and University of Texas Graduate School of Biomedical Sciences at Houston, Texas, USA.	Because interferon-alpha promotes the terminal differentiation of histiocytes and dendritic cells, we hypothesized that this molecule would be a useful therapy for Erdheim-Chester disease. We therefore treated 3 patients with advanced disease with interferon-alpha at a starting dose of 3 to 6 x 10(6) units, which was later reduced, during maintenance, to 1 x 10(6) units subcutaneous 3 times per week. Marked improvement was noted in all patients, with substantial retro-orbital disease regression within 1 month. Improvement in bone lesions, pain, diabetes insipidus, and other manifestations was gradual over many months. Responses were durable (3+ to 4.5+ years). Our observations suggest that this well-tolerated therapy has a significant effect on the course and outcome of Erdheim-Chester disease.	16020507
2005	Breast J	Erdheim-Chester disease of the breast: a case report and review of the literature.	Barnes PJ, Foyle A, Haché KA, Langley RG, Burrell S, Juskevicius R	Division of Anatomical Pathology, Queen Elizabeth II Health Sciences Center, Halifax, Nova Scotia, Canada. Penny.Barnes@cdha.nshealth.ca	We report the case of a 49-year-old woman who presented with palpable breast nodules, followed by progressive soft tissue and subcutaneous disease, and involvement of the long bones, dysarthria, and dysphagia. The histopathologic features and skeletal radiography findings are consistent with ECD. This case represents an unusual presentation, which led to delayed diagnosis, as ECD of the breast has been rarely reported. ECD should be considered in the differential diagnosis of histiocytoid breast lesions, including fat necrosis and histiocytoid invasive mammary carcinoma.	16297093
2005 Aug.	J Oral Pathol Med	Erdheim-Chester disease in a child presenting with multiple jaw lesions.	Nagatsuka H, Han PP, Taguchi K, Tsujigiwa H, Gunduz M, Fukunaga J, Sugahara T, Asaumi J, Nagai N	Department of Oral Pathology and Medicine, Graduate School of Medicine and Dentistry, Okayama University, Okayama, Japan.	We report a case of 13-year-old female patient who first presented with multiple osteolytic lesions of the jaws followed by bilateral symmetrical bone lesions affecting the lower extremities, as well as brain and abdominal involvement. Histological findings of the jaw lesions showed lipid-storing CD68 (+), CD1a (-) histiocytes with Touton type giant cells. CONCLUSION: To the best of our knowledge, this is the first case of Erdheim-Chester disease with jaw bone lesions occurring as initial presenting symptom.	16011611

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2005 Jun	Respirology	Pulmonary involvement in Erdheim-Chester disease.	Chung JH, Park MS, Shin DH, Choe KO, Kim SK, Chang J, Kim SK, Kim YS	Department of Internal Medicine, Kwandong University College of Medicine, Myungji Hospital, Koyang, Korea.	Case of a 53-year-old woman with extensive and progressive pulmonary disease. Computed tomography scans revealed diffuse infiltrative lung disease. Thoracoscopic lung biopsy and a biopsy of the right femur lesion were performed. The histopathology revealed that she had non-Langerhans' cell histiocytosis; Erdheim-Chester disease. The characteristic lesions of Erdheim-Chester disease, including involvement of the orbit, pericardium, periaorta, and bone were detected. This helped to further confirm that the patient had Erdheim-Chester disease with associated pulmonary involvement. As Erdheim-Chester disease is a rare non-Langerhans' cell histiocytosis that may be misdiagnosed as interstitial lung disease or other pulmonary disorders, this diagnosis should be considered in the differential diagnosis of such lung lesions.	15955155
2005 May	Arch Phys Med Rehabil	Erdheim-Chester disease: the effect of bisphosphonate treatment--a case report.	Eyigör S, Kirazli Y, Memis A, Başdemir G	Department of Physical Therapy and Rehabilitation, Ege University Medical Faculty, Izmir, Turkey.	We present a patient in her early sixties with bilateral mild knee and leg pain. The patient showed a typical bilateral symmetric medullary sclerosis at the diametaphyseal portions of long bones of the lower extremity. The diagnosis was confirmed by a bone biopsy, and bisphosphonate (alendronate, 70 mg/wk) was given to the patient. After 9 months of treatment, biochemical markers of bone turnover, which were high at baseline, decreased to normal ranges. However, the radiographs showed that bone lesions had changed to lytic lesions. We propose use of bisphosphonates, such as alendronate, to decrease the biochemical markers of bone turnover. But we suggest that it is premature to conclude that bisphosphonates have any effect on lytic lesions and the progression of the disease as shown by changes in radiographs. Further studies with long-term follow-up and ultrastructural evaluation are needed.	15895357
2005 May	Skeletal Radiol	Erdheim-Chester disease in a child with MR imaging showing regression of marrow changes.	Joo CU, Go YS, Kim IH, Kim CS, Lee SY	Department of Pediatrics, Chonbuk National University Medical School, 561-712 Jeonbuk, Korea.	We report a case of Erdheim-Chester disease in a 10-year-old girl evaluated with MR imaging. Radiographs revealed typical bilateral, symmetric osteosclerosis of the metaphyseal regions of long bones of the upper and lower extremities. RESULTS: A histologic examination demonstrated foamy histiocytes in bone marrow smears. Bilateral symmetric low signal intensities of both proximal tibiae and distal femurs were demonstrated on T1-weighted MR images. After oral steroid therapy for 8 months, follow-up MR imaging showed remarkable restoration of normal high signal intensity in both the tibial and femoral metaphyses. CONCLUSION: To our knowledge, this may be the first case of Erdheim-Chester disease that showed normal restoration of the abnormal signal intensities in the metaphyses of long bones after steroid therapy.	15480644
2005 Apr	Mayo Clin Proc	Laparoscopic biopsy and ureterolysis in Erdheim-Chester disease.	Castle EP, Humphreys MR, Andrews PE	Department of Urology, Mayo Clinic College of Medicine, Scottsdale, Ariz 85259, USA.	We describe a patient who underwent laparoscopic bilateral ureterolysis and laparoscopic biopsy for presumed retroperitoneal fibrosis confirmed previously by percutaneous needle biopsy findings. The final pathologic diagnosis based on laparoscopic biopsy results was ECD. As evidenced by this case, ureterolysis offers little benefit to patients with ECD.	15819294
2005 Apr	Urology	Compression of kidneys in Erdheim-Chester disease of retroperitoneum: Open surgical approach.	Wimpissinger TF, Schernthaner G, Feichtinger H, Stackl W	Department of Urology and Ludwig Boltzmann Institute for Extracorporeal Lithotripsy and Endourology, Rudolfstiftung Hospital, Vienna, Austria. florian.wimpissinger@gmx.at	We report the first case of surgical treatment of severe compression of renal parenchyma by retroperitoneal masses in a 61-year-old male patient with progressing renal failure. After 3 years of follow-up, we have concluded that the open surgical approach is an option in the management of renal complications in Erdheim-Chester disease.	15833540

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2005 Feb	Arch Pathol Lab Med	Fulminant multisystem non-langerhans cell histiocytic proliferation with hemophagocytosis: a variant form of Erdheim-Chester disease.	Rao RN, Chang CC, Uysal N, Presberg K, Shidham VB, Tomashefski JF	Department of Pathology, Medical College of Wisconsin, Milwaukee, USA.	Hemophagocytosis (HP), a feature seen in malignant histiocytosis and infection- and lymphoma-associated disorders, has not been previously emphasized in Erdheim-Chester disease (ECD). Generally, ECD is recognized as a rare, systemic, non-Langerhans cell histiocytosis with a variable clinical course. Herein, we describe a unique case of multisystem non-Langerhans cell histiocytic proliferation with a fulminant clinical course (death occurred within 3 months of presentation) that showed prominent HP and extensive involvement of multiple organs, including the lungs, resulting in respiratory failure. Hemophagocytosis led to severe anemia that required transfusion and thrombocytopenia. Antemortem lung and bone marrow biopsy specimens revealed involvement by a histiocytic infiltrate with features highly suggestive of ECD and HP. Furthermore, the autopsy documented the presence of HP and the histiocytic infiltrate in multiple other organs. This case is best categorized as a variant form of ECD. Recognizing this variant has the following important implications: (1) HP may be a marker for fulminant clinical course in ECD, (2) the presence of HP does not exclude a diagnosis of ECD, and (3) ECD should be considered in the differential diagnosis of HP.	15679446
2005 Jan	Dtsch Med Wochenschr	[Erdheim-Chester disease]	**Koziolek MJ, Kunze E, Müller A, Thiem V, Scheel AK, Müller D, Müller GA, Strutz F**	Abteilung Nephrologie und Rheumatologie, Georg-August-Universität Göttingen. mkoziolek@gmx.de	A 55-year-old female was admitted complaining of musculoskeletal pain and weakness of both lower extremities for a number of years. Due to a hypothalamic mass of unknown aetiology a diabetes insipidus, a gonadotrophic, somatotrophic and a partially corticotrophic insufficiency had developed. Investigations indicated Erdheim-Chester disease (ECD). Under treatment with glucocorticosteroids musculoskeletal complaints improved, but re-appeared following dose reduction. A therapeutic trial using methotrexat did not affect the complaints. The Erdheim-Chester syndrome is considered to belong to diseases with a proliferation of the monocytic-histiocytic and dendritic cellular system. In the presence of symmetric musculoskeletal symptoms associated with osteosclerotic and osteolytic lesions particularly occurring in the long bones of the lower extremities and concomitant with elevated serum markers of inflammation, the Erdheim-Chester disease should be taken into account. To date, no validated therapy exists.	15619170
2005	Int Urol Nephrol	Renal calculi in a patient with Erdheim-Chester disease.	Dundee P, Bouchier-Hayes D, Iles L, Costello A	Department of Urology, Royal Melbourne Hospital, Parkville, Melbourne, Australia. pdundee@amavic.com	We report a patient with long standing ECD with widespread extraskeletal involvement, including significant renal infiltration, presenting with left hydronephrosis secondery obstruction from a proximal ureteric calculas.	16307316
2004 Dec	An Med Interna	[Erdheim-Chester disease and Langerhans histiocytosis. A fortuitous association?]	Simiele Narvarte A, Novoa Sanjurjo F, Gómez Rodríguez N, Antón Badiola I	Servicio de Hematología, Centro Médico POVISA, Vigo, Pontevedra.	We present a new case with histological data of both histiocytosis whose clinical course included bone and muscle pain, insipidus diabetes, exophthalmos, bilateral symmetrical sclerosing bone lesions and a cerebellar syndrome.	15628955

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2004 Dec	Arch Pathol Lab Med	Pulmonary and ophthalmic involvement with Erdheim-Chester disease: a case report and review of the literature.	Allen TC, Chevez-Barrios P, Shetlar DJ, Cagle PT	Department of Pathology, University of Texas Health Center at Tyler, Houston, Tex 75708-3154, USA. Timothy.Allen@uthct.edu	Erdheim-Chester disease is a rare nonfamilial histiocytic disorder of unknown etiology with characteristic long bone findings. The 3-year survival rate for patients with Erdheim-Chester disease is 50%. Approximately 50% of patients have disease involvement in other tissues, including skin, retro-orbital and periorbital tissues, pituitary-hypothalamic axis, heart, kidney, retroperitoneum, breast, skeletal muscle, and sinonasal mucosa; about 20% of patients have lung involvement. Prognosis generally depends on the extent of the extraosseous disease. For patients with lung involvement, gender distribution is equal, but men typically present at an older age than do women. Approximately 80% of patients present with dyspnea, and most patients have diffuse interstitial infiltrates and pleural and/or interlobar septal thickening on chest radiology. Characteristic lung histopathology includes the accumulation of histiocytes with variable amounts of fibrosis and a variable lymphoplasmacytic infiltrate in a lymphangitic distribution. Immunostains are diagnostically useful, showing immunopositivity for CD68 and factor XIIIa and immunonegativity for CD1a. Birbeck granules are uniformly absent ultrastructurally.	15578889
2004 Dec	Clin Radiol	Erdheim-Chester disease versus multifocal fibrosis and Ormond's disease: a diagnostic dilemma.	Bangard C, Lotz J, Rosenthal H, Galanski M	Department of Radiology, University of Cologne, Cologne, Germany. cbangard@gmx.de	The aim of the study was to evaluate the effectiveness of different imaging techniques with respect to diagnosis and differential diagnosis between Erdheim-Chester disease (ECD) and multifocal fibrosis (MF)/Ormond's disease (OD). METHOD: Three cases of ECD were included, two of which were misdiagnosed as MF/OD. Findings in different imaging techniques [plain radiography, skeletal scintigraphy, computed tomography (CT) and magnetic resonance imaging (MRI)] of the lower extremities, chest MRI, craniofacial MRI, abdominal CT and MRI) were compared and ranked with regard to diagnostic efficacy. RESULTS: Differentiation between ECD and MF/OD is only possible by imaging the long bones. Bone roentgenograms and skeletal scintigraphy, followed by MRI and CT of the lower extremities are the most effective imaging techniques. CONCLUSION: A low threshold for carrying out plain radiography of the lower limbs in case of RF/MF will increase the number of ECD-cases.	15556598
2004 Dec	Diagn Cytopathol	Erdheim-Chester disease of the brain: cytological features and differential diagnosis of a challenging case.	Rushing EJ, Kaplan KJ, Mena H, Sandberg GD, Koeller K, Bouffard JP	Department of Neuropathology and Ophthalmic Pathology, Armed Forces Institute of Pathology, Washington, DC 20306-6000, USA. rushinge@afip.osd.mil	We describe a case that presented in the brain of a 26-yr-old male patient and clinically mimicked the appearance of a neoplasm. The final diagnosis was a surprise. In retrospect, the diagnosis was suggested by the intraoperative "squash" preparations, which demonstrated a mixed cellular proliferation of lymphohistiocytic elements and large, multinucleated cells with vesicular nuclei, prominent nucleoli, and abundant cytoplasm. To the best of our knowledge, this is the first report detailing the cytopathological features of ECD.	15540182
2004 Nov	J Clin Pathol	Erdheim-Chester disease: case report with multisystemic manifestations including testes, thyroid, and lymph nodes, and a review of literature.	Sheu SY, Wenzel RR, Kersting C, Merten R, Otterbach F, Schmid KW	Institute of Pathology, University of Essen, 45122 Essen, Germany. sein-yi.sheu@medizin.uni-essen.de	This report describes the case of a 50 year old white man who presented with hypogonadism and diabetes insipidus. At necropsy, extensive organ involvement was found, including the testes, thyroid, and lymph nodes. This is the first report of thyroid and lymph node infiltration in this disease. Because of the endocrinological symptoms, neurosarcoidosis and hypophysitis are important diseases in the differential diagnosis. This report also includes a review of the literature concerning rare organ manifestations and patients presenting primarily with similar symptoms.	15509691

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2004 Nov	Klin Monatsbl Augenheilkd	[Erdheim-Chester disease as differential diagnosis in bilateral exophthalmos]	Röpke E, Herde J, Bloching M	Klinik und Poliklinik für Hals-, Nasen- und Ohrenheilkunde, Kopf- und Halschirurgie, Halle. ernst.roepke@medizin.uni-halle.de	This report describes the case of a patient who had symmetrical exophthalmos, periorbital xanthelasmas and reduced vision. Next to Wegener's granulomatosis the differential diagnosis of Erdheim-Chester disease is discussed. It concerns a rare systemic histiocytosis of unknown etiology. Above all, the skeleton system with symmetrical long bone osteosclerosis is affected. Manifestations in the area of the orbit have seldom been reported with bilateral retrobulbar infiltrations, exophthalmos, diplopia, compression of the optic nerve and periorbital xanthelasmas.	15562361
2004 Nov	Medicine (Baltimore)	Cardiovascular involvement, an overlooked feature of Erdheim-Chester disease: report of 6 new cases and a literature review.	Haroche J, Amoura Z, Dion E, Wechsler B, Costedoat-Chalumeau N, Cacoub P, Isnard R, Généreau T, Wechsler J, Weber N, Graef C, Cluzel P, Grenier P, Piette JC	Service de Médecine Interne, Hôpital Pitié-Salpêtrière, Paris, France. julien.haroche@psl.ap-hop-paris.fr	Cardiovascular manifestations of ECD remain underestimated. We report 6 new cases of ECD associated with periaortic fibrosis. In 4 of these cases, the whole aorta had a "coated" aspect. A literature review revealed 66 cases of ECD with cardiovascular involvement. We therefore analyzed 72 ECD patients with cardiovascular involvement. Data concerning follow-up were available for 58 (80.6%) patients. Of these, 35 (60.3%) patients died, confirming the severe prognosis of ECD. Cardiovascular complications were responsible for the death of 11 of the 35 patients (31.4%).	15525849
2004 Oct	Arch Soc Esp Oftalmol	[Orbit xanthogranulomatosis. Erdheim-Chester disease]	Rozas Reyes P, Señaris González A, González Rodríguez CM	Hospital Universitario Central de Asturias, Spain. prozas@telecable.es	A patient was studied because of upper lid bilateral edema and xanthelasmae-like lesions after three years of evolution. During the ophthalmologic examination orange-yellowish lesions and two symmetrical tumours were observed on the temporal part of both upper lids. Corticoid-therapy was undertaken which reduced the size of the tumours, however the size increased again after the discontinuation of treatment. A biopsy was performed and lid xanthogranulomatosis was diagnosed. Other systemic examinations were normal. DISCUSSION: Erdheim-Chester disease is a xanthogranulomatosis that can affect ocular and periorbital structures. Combination of xanthelasmae-like lesions and bilateral orbital masses should make us consider this process and try to locate any associated systemic conditions.	15523574
2004 Oct	Headache	Familial hemiplegic migraine, neuropsychiatric symptoms, and Erdheim-Chester disease.	Black DF, Kung S, Sola CL, Bostwick MJ, Swanson JW	Mayo Clinic, Neurology, Rochester, MN 55905, USA.	We report the occurrence of unilateral cerebral hemisphere edema with subsequent cortical laminar necrosis in the setting of familial hemiplegic migraine (FHM) and permanent neurologic sequelae after resolution of an attack in 1 patient. Contemporaneous with this severe attack of FHM, the patient was found to exhibit multiple systemic and neurological symptoms referable to Erdheim-Chester disease (a rare non-Langerhans cell histiocytosis) that was confirmed by bone biopsy. This case demonstrates the severity possible with a migrainous infarction associated with FHM. The co-occurrence of two such rare entities in 1 patient suggests a possible relationship.	15447701
2004 Oct	Virchows Arch	Erdheim-Chester disease of the breast associated with Langerhans-cell histiocytosis of the hard palate.	Andrade VP, Nemer CC, Prezotti AN, Goulart WS	Fleury, Centro de Medicina Diagnóstica, Av. Gal Waldomiro de Lima, 508. Jabaquara, CEP 04344-070, Saõ Paulo, Brazil. victor.andrade@fleury.com.br	We report a patient with Langerhans-cell histiocytosis (LCH) localized to the hard palate that was later proven to be associated with Erdheim-Chester disease (ECD), involving the right breast, skeleton, retroperitoneum and left orbit. Mammary involvement by ECD is an extremely rare condition, which should be differentiated from some benign and malignant mimickers, especially the histiocytoid type of breast carcinoma. Characteristic histological features plus clinical and radiographic information are needed to achieve a correct diagnosis. The ECD, its relation to the LCH and details of the breast lesion are discussed.	15338304

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2008 Apr	61: Eur Heart J;	A rare cause of cardiac tumour: an Erdheim-Chester disease with cardiac involvement co-existing with an intracerebral Langerhans cell histiocytosis.	Granier M, Micheau A, Serre I	Department of Cardiology, Arnaud de Villeneuve, Avenue du Doyen G Giraud, Montpellier 34000, France.		18390872
2007 Jun	62: Ann Hematol;	Erdheim-Chester disease with hemophagocytosis.	Busemann C, Kallinich B, Schwesinger G, Krüger W, Schüler F, Schmidt CA, Dölken G	Department of Hematology and Oncology, University Medical Center, Ernst-Moritz-Arndt-University Greifswald, Sauerbruchstraße, 17487, Greifswald, Germany, busemann@uni-greifswald.de.		17579863
2007 Jun	63: Br J Haematol;	Multisystem Erdheim-Chester disease; a unique presentation with liver and axial skeletal involvement.	Gupta A, Aman K, Al-Babtain M, Al-Wazzan H, Morouf R	Department of Haematology, Mubarak Al- Kabeer hospital, Faculty of Medicine, Kuwait University, Jabriya, Kuwait.		17553060
2007 Jul	64: Arch Dermatol;143(7):952-3	Verruca plana-like papules as a new manifestation of erdheim-chester disease.	Yanagi T, Kato N, Yamane N, Osawa R, Hiraga H	Department of Dermatology, National Hospital Organization Hokkaido Cancer Center, Kikusui 4-2, Shiroishi-ku, 003-0804, Sapporo, Japan. yanagi@med.hokudai.ac.jp.		17638752
2007 Jun	65: Hum Pathol;38(6):950-1	Intracranial lesion of Erdheim-Chester disease.	Shimada S, Ono K, Hashizume Y, Nakaguro M, Suzuki Y, Mori N	Currently, Department of Pathology and Clinical Laboratories/Nagoya University Hospital Previously, Department of Pathology of Biological Response, Nagoya University Graduate School of Medicine, Nagoya 466-8550, Japan.		17509397

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2007 Mar	66: Nephrol Dial Transplant;	Erdheim-Chester disease: a rare cause of renal failure.	Verdalles U, Goicoechea M, García de Vinuesa S, Mosse A, Luño J	Department of Nephrology, Hospital General Universitario Gregorio Marañon, Spain.		17395655
2007 Apr	67: Circulation;115(16):e412-4	Images in cardiovascular medicine. Magnetic resonance imaging guiding pacemaker implantation for severe sinus node dysfunction due to cardiac involvement in Erdheim-Chester disease.	Elgeti T, Schlegl M, Nitardy A, Kivelitz DE, Stockburger M	Department of Radiology, Charité-Universitätsmedzin Berlin, Charitéplatz 1, 10117 Berlin, Germany. thomas.elgeti@charite.de		17452611
2007 Feb	68: J Am Coll Surg;204(2):326-7	Mesenteric panniculitis and Erdheim-Chester disease: xanthogranulomatous diseases confused with malignancy.	Moore FO, Berne JD, Fox AD	East Texas Medical Center, Tyler, TX, USA.		17254937
2007 Jan	69: Clin Nucl Med;32(1):35-8	Tc-99m MDP bone scintigraphy and positron emission tomography/computed tomography (PET/CT) imaging in Erdheim-Chester disease.	Namwongprom S, Núñez R, Kim EE, Macapinlac HA	Department of Nuclear Medicine, The University of Texas M.D. Anderson Cancer Center, Houston, Texas 77030, USA. snamwong@mail.med.cmu.ac.th		17179801
2006 Nov	70: J Nucl Cardiol;13(6):867-9	Dramatic change of Ga-67 citrate uptake before and after corticosteroid therapy in a case of cardiac histiocytosis (Erdheim-Chester disease).	Kudo Y, Iguchi N, Sumiyoshi T, Murai T, Oka T	Department of Cardiovascular Internal Medicine, Sakakibara Heart Institute, Tokyo, Japan. yoko-kd@kd5.so-net.ne.jp		17174817
2006 Sep	71: J Neurol Neurosurg Psychiatry;77(9):1078	Neurological picture. Torcular Erdheim-Chester disease.	Gazzeri R, Galarza M, Amoroso R, De Bonis C, D'Angelo V	Department of Neurosurgery, San Giovanni Addolorata Hospital, Rome, Italy. robertogazzeri@gmail.com		16914757

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2006 May	72: Leuk Lymphoma;47(5):935-7	18F-FDG positron emission tomographic imaging in Erdheim-Chester disease with skeletal and extra-skeletal involvement.	Nakahara T, Suzuki T, Uno K, Joishi D, Tanaka C, Hashimoto J, Kubo A			16753885
2006 Feb	73: Clin Nucl Med;31(2):90-2	Tc-99m MDP bone and Ga-67 citrate scintigraphy of Erdheim-Chester disease in a child.	Sohn MH, Kim MW, Kang YH, Jeong HJ	Department of Nuclear Medicine, Chonbuk National University, Chonbuk, Korea. mhsohn@chonbuk.ac.kr		16424695
2005	74: Rev Esp Med Nucl;24(6):423	[Occult femoral neck fracture in a patient with Erdheim-Chester disease]	Ramos-Font C, Rebollo Aguirre AC, Moral Ruiz A, Bellón Guardia M, Cabello García D, Llamas-Elvira JM	Servicio de Medicina Nuclear, Hospital Universitario Virgen de las Nieves, Granada. cramof@fundacionhvn.org		16324522
2005 Aug	75: Eur J Nucl Med Mol Imaging;32(8):998	Erdheim-Chester disease: 99mTc-MDP bone scan provides the diagnosis.	Canbaz F, Dabak N, Baris S, Selcuk MB	Department of Nuclear Medicine, Ondokuz Mayis University, Samsun, Turkey.		15864582
2005 Aug	76: J R Soc Med x;98(6):296	Erdheim-Chester disease.	Rhodes B, Jawad AS			15928398
2005 May	77: J Radiol;86(5 Pt 1):527-30	[Case #5. Erdheim-Chester disease]	Cattin F, Runge M, Magy N, Dupont JL, Bonneville JF	Service de Neuroradiologie, CHU - Hôpital Jean-Minjoz, Boulevard Alexander-Fleming, 25030 Besançon.		16114215
2005 Jan	78: Clin Nucl Med;30(1):32-4	Radionuclide bone imaging in Erdheim-Chester disease.	Núñez R, Tronco GG, Rini JN, Hofman J, Amoashiy M, Bhuiya T, Palestro CJ	Division of Nuclear Medicine, Department of Radiology, Long Island Jewish Medical Center, New Hyde Park, New York, USA. rodolfo.nunez@di.mdacc.tmc.edu		15604967

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2005 Jan	79: Radiology;234(1):111-5	Case 78: Erdheim-Chester disease with central nervous system involvement.	Adem C, Hélie O, Lévêque C, Taillia H, Cordoliani YS	Department of Radiology, Hôpital d'Instruction des Armées du Val-de-Grâce, 74 boulevard de Port Royal, F-75005 Paris, France. valimage@wanadoo.fr		15618378
2004 Nov	80: AJR Am J Roentgenol;183(5):1253-60	Imaging of thoracoabdominal involvement in Erdheim-Chester disease.	Dion E, Graef C, Haroche J, Renard-Penna R, Cluzel P, Wechsler B, Piette JC, Grenier PA	Department of Radiology, Hôpital Pitié-Salpêtrière, Assistance Publique-Hôpitaux de Paris-Université Pierre-et-Marie-Curie, 47-83 Boulevard de l'Hôpital, 75651 Paris Cedex 13, France.		15505288
2004 Oct	81: Circulation;110(15):e443-4	Images in cardiovascular medicine. High resolution images obtained with ultrasound and magnetic resonance imaging of pericarotid fibrosis in Erdheim-Chester disease.	Gauvrit JY, Oppenheim C, Girot M, Lambert M, Gautier C, Hatron PY, Pruvo JP, Leclerc X	Department of Neuroradiology and EA 2691, University Hospital of Lille, Lille, France. jygauvrit@chru-lille.fr		15477423
2008 Feb	82: J Thorac Imaging;23(1):7-12	CT-guided Biopsy of Nonresolving Focal Air Space Consolidation.	Ferretti GR, Jankowski A, Rodière M, Brichon PY, Brambilla C, Lantuejoul S	*Service Central de Radiologie et Imagerie Médicale ‡Département de Chriurgie Thoracique et Vasculaire ‖ Département de Pathologie Cellulaire, INSERM U 823 †Service Central de Radiologie et Imagerie Médicale, CHU Grenoble §INSERM U 823, Institut A Bonniot, Grenoble, France.	OBJECTIVES: To evaluate the diagnostic accuracy of percutaneous computed tomography (CT)-guided coaxial core needle biopsy in patients with nonresolving pulmonary focal air space consolidations and negative fiberoptic bronchoscopy results. METHODS: From 1997 to 2005, 23 patients (11 woman, 12 men; age range, 45 to 81 y; mean age, 66 y) presenting with nonresolving pneumonia persisting more than 8 weeks (mean, 22 wk; range, 8 to 40 wk) with negative fiberscopic results, underwent coaxial percutaneous biopsy using an automated core needle (18-gauge) under CT guidance. Histologic and bacteriologic evaluations were obtained. The final diagnosis was confirmed by surgical pathology, culture results, or clinical follow-up. RESULTS: Specimens adequate for histopathologic evaluations were obtained in 20 (87%) cases. Final diagnoses were lung cancer (n=15) and benign diseases (infectious pneumonia, 3; lipoid pneumonia, 1; Erdheim Chester disease: 1; and nonspecific chronic pneumonia, 3). Diagnostic yield of core needle biopsy was 78% (18 of 23). The sensitivity and specificity for malignancy were 87% and 100%, respectively. Immediate pneumothorax was present in 11 patients of cases, but only 2 patients required pleural drainage. DISCUSSION: CT-guided lung biopsy using a core needle biopsy provides a high degree of diagnostic accuracy and allows specific characterization of nonresolving pulmonary focal air space consolidation.	18347513

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2007 Aug	83: Semin Diagn Pathol;24(3):162-82	Histiocytic lesions and proliferations in the lung.	Wang CW, Colby TV	Department of Pathology, Mayo Clinic College of Medicine, Scottsdale, Arizona 85259, USA.	Pulmonary lesions encountered by the pathologist in which histiocytes are the dominant finding histologically are reviewed. Lesions discussed include neoplasms of histiocytes and nonneoplastic processes. Entities of uncertain histogenesis, including Rosai-Dorfman disease and Erdheim-Chester disease, are also discussed. Qualitative features of the histiocytes are addressed, including the presence of foreign dust, hemosiderin, foamy change, and histiocytes showing features of Langerhans' cells.	17882900
2006 Sep	84: Orbit;25(3):221-5	Orbital and eyelid manifestations of xanthogranulomatous diseases.	Vick VL, Wilson MW, Fleming JC, Haik BG	Department of Ophthalmology, University of Tennessee Health Science Center, Memphis, Tennessee, USA.	Erdheim-Chester disease, adult periocular xanthogranuloma, juvenile xanthogranuloma, and necrobiotic xanthogranuloma are presumed to be separate disease entities, but they are often confused clinically because of their similar presentations and histopathology. To further describe the xanthogranulomatous diseases and to identify possible pitfalls in their diagnoses, we retrospectively reviewed charts from 1998 to 2001 for all patients with biopsy-proven xanthogranulomatous process of the eyelid and/or orbit. We found 2 patients diagnosed with adult periocular xanthogranuloma and 1 with Erdheim-Chester disease, each case initially misdiagnosed. Careful review of the clinical manifestations, histopathological review of all previous biopsy specimens, and repeat biopsy aided in the correct diagnosis and management of disease in these 3 patients.	16987770
2006 Jun	85: J Fr Ophtalmol;29(6):672-86	[Pathology of the eyelid in elderly patients]	Thomas L, Dalle S	Service de Dermatologie, Hôtel Dieu, Lyon. luc.thomas@chu-lyon.fr	Pathology of the eyelids in elderly patients is extremely polymorphic. It is mainly centered on skin cancers (basal cell carcinoma, squamous cell carcinoma, adnexal carcinomas, and melanoma). Most severe aspects of the inflammatory diseases of the eyelid are bullous diseases (cicatricial pemphigoid, pemphigus, Stevens-Johnson syndrome, etc.). A number of rare diseases deserve mention since their presence could lead to the diagnosis of internal or systemic diseases (dermatomyositis, necrobiotic xanthogranuloma, Erdheim-Chester, etc.). In such conditions, early diagnosis is often based on the observation of isolated periocular symptoms. CONCLUSIONS: Even though topographic dermatology is a somewhat reductive vision of skin diseases, pathology of the eyelids deserves special mention because of its polymorphism as well as its diagnostic and/or therapeutic significance.	16885900
2006 May	86: Br J Ophthalmol;90(5):602-8	Adult xanthogranulomatous disease of the orbit and ocular adnexa: new immunohistochemical findings and clinical review.	Sivak-Callcott JA, Rootman J, Rasmussen SL, Nugent RA, White VA, Paridaens D, Currie Z, Rose G, Clark B, McNab AA, Buffam FV, Neigel JM, Kazim M	Department of Ophthalmology, West Virginia University Eye Institute, Morgantown, 26505, USA. jsivak@hsc.wvu.edu	BACKGROUND/AIMS: Adult xanthogranulomatous disease involving the ocular tissues is rare and poorly understood. Adult onset xanthogranuloma (AOX), adult onset asthma and periocular xanthogranuloma (AAPOX), necrobiotic xanthogranuloma (NBX), and Erdheim-Chester disease (ECD) are the four syndromes within this disorder, which is diagnosed by characteristic histopathology. 22 cases, including histopathological slides, were compiled. 137 cases were compiled. Adult xanthogranuloma of the orbit is rare, making prospective evaluation or meta-analysis impossible. The best treatment is unknown but seems to be with multiagent chemotherapy guided by histopathological, immunohistochemical, and systemic findings.	16622091
2006 Mar	87: Neth J Med;64(3):88-90	Pleural thickening in a construction worker: it is not always mesothelioma.	Saboerali MD, Koolen MG, Noorduyn LA, van Delden OM, Bogaard HJ	Department of Respiratory Medicine, Academic Medical Centre, Amsterdam, the Netherlands.	We describe the case of a 45-year-old man presenting with chest pain and pleural effusions. These symptoms were progressive over a period of three years, with pericardial involvement and respiratory insufficiency finally resulting in death. Despite repeated diagnostic procedures, a final diagnosis could only be made at autopsy. Multisystem foamy histiocyte infiltration suggested the diagnosis of Erdheim-Chester disease.	16547363

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2006 Feb	88: Rheumatology (Oxford);45(2):192-5	Diagnostic value of blind synovial biopsy in clinical practice.	Kroot EJ, Weel AE, Hazes JM, Zondervan PE, Heijboer MP, van Daele PL, Dolhain RJ	Erasmus MC, Department of Rheumatology, Z-712, PO Box 2040, 3000 CA Rotterdam, The Netherlands.	OBJECTIVE: To assess the diagnostic value of blindly performed synovial biopsies in carefully selected patients with unclassified arthritis. METHODS: Synovial tissue was Four patients with unclassified arthritis could be diagnosed properly based upon examination of synovial tissue of the knee obtained by an easy-to-perform blind biopsy. The arthritis of the four patients was diagnosed as being part of Erdheim-Chester disease, sarcoidosis, multicentric reticulohistiocytosis and arthritis caused by foreign-body material, respectively. CONCLUSIONS: Analysis of synovial tissue obtained during a blind biopsy procedure has diagnostic potential in carefully selected patients with unclassified arthritis. The common denominator in all the cases presented was a differential diagnosis consisting of a rheumatological disease with characteristic histological features.	16234280
2005	89: Radiographics; 25(3):719-30	Unusual nonneoplastic peritoneal and subperitoneal conditions: CT findings.	Pickhardt PJ, Bhalla S	Department of Radiology, University of Wisconsin Medical School, Madison, WI 53792, USA. ppickhardt@mail.radiology.wisc.edu	Peritoneal disease can manifest at computed tomography (CT) as fluid accumulation within the peritoneal cavity (ascites) or soft-tissue infiltration of the various peritoneal ligaments and mesenteries. Beyond the commonly encountered cases of typical ascites and peritonitis, there is a wide spectrum of uncommon nonneoplastic conditions that may involve the peritoneal and subperitoneal spaces. For example, systemic or organ-based diseases that occasionally involve the peritoneum include eosinophilic gastroenteritis, amyloidosis, extramedullary hematopoiesis, Erdheim-Chester disease, sarcoidosis, and mesenteric cavitary lymph node syndrome. Tumorlike conditions that may affect the peritoneum include aggressive fibromatosis (desmoid), inflammatory pseudotumor, retractile mesenteritis, and Castleman disease. Atypical peritoneal infections include tuberculosis, actinomycosis, echinococcosis, Whipple disease, and mesenteric adenitis. Conditions involving the subperitoneal fat include epiploic appendagitis, mesenteric panniculitis, and segmental omental infarction, all of which have characteristic CT findings. CT is an excellent imaging modality for detection and characterization of peritoneal involvement from these unusual diseases.	15888621
2005 Mar	90: J Vasc Surg;41(3):457-61	Use of the ascending aorta as bypass inflow for treatment of chronic intestinal ischemia.	Chiche L, Kieffer E	Department of Vascular Surgery, Pitié-Salpêtriére University Hospital, 47-83 boulevard de l'Hôpital, Assistance Publique-Hopitaux de Paris, Paris, France.laurent.chiche@psl.ap-hop-paris.fr	In this report, we describe our experience with an antegrade bypass technique from the ascending aorta in patients with no other available inflow. METHODS: From April 1990 to May 2004, we performed antegrade bypass from the ascending aorta to the celiac artery, superior mesenteric artery (SMA), or both in five patients. These cases accounted for 2.4% of the 211 patients who underwent surgery on intestinal arteries during the study period. Results: Four patients presented with symptomatic CII, and one patient had no intestinal ischemic symptoms. The underlying disease was Takayasu disease in two cases, Erdheim-Chester disease in one case, chronic aortic dissection in one case, and atherosclerosis in one case. CONCLUSION: Antegrade intestinal artery bypass from the ascending aorta is an effective alternative for patients who have no other available inflow for conventional antegrade or retrograde bypass and for patients in whom major technical difficulties are likely after multiple exposures of the thoracoabdominal aorta. Although indications are uncommon, antegrade intestinal artery bypass can provide durable revascularization of the intestine.	15838480

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2005 Feb	91: Clin Radiol;60(2):171-88	The dural tail sign--beyond meningioma.	Guermazi A, Lafitte F, Miaux Y, Adem C, Bonneville JF, Chiras J	Department of Radiology, University of California, San Francisco, USA. ali.guermazi@synarc.com	There have been somewhat conflicting reports published about the significance of linear meningeal thickening and enhancement adjacent to peripherally located cranial mass lesions on contrast-enhanced magnetic resonance (MR) images. Most of the authors consider this so-called "dural tail sign" or "flare sign" almost specific for meningioma. This review illustrates the MR imaging findings of a wide spectrum of disorders that show this dural sign. Causes include other extra-axial lesions and also peripherally located intra-axial lesions such as neuromas, chloromas, metastases, lymphoma, gliomas, pituitary diseases, granulomatous disorders, and also cerebral Erdheim-Chester disease. The dural tail sign is not specific to a particular pathological process. Nevertheless, useful conclusions can be drawn from the morphology of the lesion, its enhancement pattern, and its solitary or multifocal presentation. The final diagnosis must be based on cerebrospinal fluid studies or histological studies after biopsy.	15664571
2004 Nov	92: J Neurosurg;101(5):864-8	Preoperative stent placement for intradural vertebral artery stenosis from a rare xanthogranuloma. Case report.	Boulos AS, Deshaies EM, Qian J, Popp AJ	Department of Surgery, Division of Neurosurgery Albany Medical Center, Albany, New York 12208, USA. boulosa@mail.amc.edu	In this report, the authors discuss a novel use of intradural vertebral artery (VA) stent placement to protect a tumor-encased vessel from injury during lesion resection. The tumor was a rare foramen magnum region xanthogranuloma and a component of Erdheim-Chester disease (ECD). This 64-year-old man presented with large masses encasing and compressing the intracranial segments of each VA. Preoperatively, a left VA stent was placed to protect the arterial wall during resection of the tumor. Histopathological study results on the subtotally resected mass were consistent with xanthogranuloma, a rare benign histiocytic tumor frequently occurring in patients with ECD. Further radiographic evaluation in the patient revealed an osteolytic lesion of the eleventh thoracic vertebra supporting the diagnosis of ECD disease. Based on this case study, the authors recommend the following: 1) tumor-encased vessels can be protected preoperatively by stent placement to assist with tumor debulking; and 2) patients diagnosed with a xanthogranuloma should be evaluated for multisystem involvement consistent with ECD.	15540929
2007 Mar	94: Br J Radiol;80(951):227-9	An unusual cause of knee pain.	Charest M, Haider EA, Rush C	Department of Nuclear Medicine, Division of Radiology, Jewish General Hospital, McGill University, Room G-19, 3755 Cote St. Catherine Road, Montreal, Quebec, H3T 1E2, Canada. charestm@myway.com		17548507
2006 Aug	95: Pathologica;98(4):211-23	[Diagnostic utility of macrophages in interstitial lung disease]	Cavazza A, Rossi G, Barbareschi M, Damiani S, Cancellieri A, Murer B	Unità Operative di Anatomia Patologica, Ospedale S. Maria Nuova di Reggio Emilia, Italy. cavazza.alberto@asmn.re.it		17175789
2006 Apr	96: Clin Orthop Relat Res;445:261-8	Bilateral lower extremity discomfort in a 64-year-old woman.	Bugnone AN, Temple HT, Humble S	Department of Radiology, University of Miami School of Medicine, Miami, Florida, USA. bugnonea@yahoo.com		16601420

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2005 Jun	97: Arch Soc Esp Oftalmol;80(6):329	[Non-Langerhans' cell histiocytosis]	E Mencía-Gutiérrez			15986272
2005 Jun	98: Arch Soc Esp Oftalmol;80(6):329-30	[Orbital xanthogranulomatosis]	Mateos G Blanco			15986271
2005 Apr	99: J R Soc Med;98(4):165-6	Renal bone disease.	Lee JH, Stodell M, Fowler JC	Department of Medicine, Luton and Dunstable NHS Trust, Lewsey Road, Luton LU4 0DZ, UK.		15805559
2005 Jan	100: AJNR Am J Neuroradiol;26(1):34-8	Intradural spinal vein enlargement in craniospinal hypotension.	Burtis MT, Ulmer JL, Miller GA, Barboli AC, Koss SA, Brown WD	Division of Neuroradiology, Department of Radiology, Medical College of Wisconsin, Milwaukee, WI 53226, USA.	We present a case of craniospinal hypotension in a 45-year-old woman with an associated epidural pseudomeningocele extending the entire length of the spine. The epidural pseudomeningocele was caused by a CSF leak at the T8 level. In addition to typical low-pressure symptoms, the epidural pseudomeningocele caused atypical symptoms characterized by positional thoracic radiculopathy. Craniospinal hypotension was associated with massive cervical epidural venous engorgement, as well as enlargement of the posterior spinal cord vein, which was reminiscent of a dural arteriovenous fistula at CT myelography. Enlargement of the posterior spinal vein is explained by the Monro-Kellie hypothesis, and the spinal analog to enlarged cerebral veins known to be associated with intracranial hypotension.	15661695
2005 May	1: Skeletal Radiol;34(5):299-302	Erdheim-Chester disease in a child with MR imaging showing regression of marrow changes.	Joo CU, Go YS, Kim IH, Kim CS, Lee SY	Department of Pediatrics, Chonbuk National University Medical School, 561-712 Jeonbuk, Korea.	We report a case of Erdheim-Chester disease in a 10-year-old girl evaluated with MR imaging. Radiographs revealed typical bilateral, symmetric osteosclerosis of the metaphyseal regions of long bones of the upper and lower extremities. RESULTS: A histologic examination demonstrated foamy histiocytes in bone marrow smears. Bilateral symmetric low signal intensities of both proximal tibiae and distal femurs were demonstrated on T1-weighted MR images. After oral steroid therapy for 8 months, follow-up MR imaging showed remarkable restoration of normal high signal intensity in both the tibial and femoral metaphyses. CONCLUSION: To our knowledge, this may be the first case of Erdheim-Chester disease that showed normal restoration of the abnormal signal intensities in the metaphyses of long bones after steroid therapy.	15480644

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2004 Sep	12: Graefes Arch Clin Exp Ophthalmol;242 (9):803-7	Erdheim-Chester disease: a case report.	Hoffmann EM, Müller-Forell W, Pitz S, Radner H	Department of Ophthalmology, University of Mainz, Langenbeckstr.1, 55131 Mainz, Germany. ehoffman@mail.uni-mainz.de	A 61-year-old man presented with bilateral exophthalmos and progressive loss of visual function caused by chorioretinal folds and papillary swelling due to retrobulbar pseudotumor. Retrobulbar radiotherapy (20 Gy) and long-term systemic corticosteroid treatment followed. Although the retroperitoneal involvement decreased, no significant effect on orbital involvement was achieved. A second review of the orbital biopsy revealed foamy cell infiltration and the presence of a sclerotic process. Immunohistochemical examination demonstrated positive CD 68 stains, whereas S-100 and CD 1a were negative, thus confirming ECD. The histologic finding was comparable to a biopsy of the retroperitoneum. Endonasal decompression was performed but visual acuity (VA) decreased to 20/250 in the right eye and on finger counting in the left eye. The patient continues to be under therapy with prednisolone 20 mg/day and methotrexate 25 mg/week. CONCLUSIONS: The clinical orbital manifestation of ECD occurs in two different forms: one presenting as a mild impairment of visual function, while the second, clinical form, observed in our patient, is characterized by a progressive loss of VA despite therapeutic efforts such as immunosuppressive systemic therapy, radiation, and surgery. The described case illustrates that clinical findings in multifocal fibrosclerosis overlap with those observed in ECD.	15221300
2004 Sep	13: J Neurosurg;101(3):521-7	Diagnosis of Erdheim-Chester disease by using computerized tomography-guided stereotactic biopsy of a caudate lesion. Case report.	Tashjian V, Doppenberg EM, Lyders E, Broaddus WC, Pavot P, Tye G, Liu AY, Perez J, Ghatak N	Department of Neurosurgery, Medical College of Virginia Hospitals, Virginia Commonwealth University, Richmond, Virginia 23298-0631, USA.	A 27-year-old woman with Erdheim-Chester disease (ECD) and extensive intracranial involvement, in whom the initial diagnosis of ECD was established based on computerized tomography (CT)-guided stereotactic biopsy of a caudate lesion. In the setting of neurological involvement, neurosurgical biopsy has been reported seven times in the literature, with only one of these biopsies being the basis for the initial diagnosis of the disease. The authors' case represents only the second time the disease has been diagnosed by means of neurosurgical biopsy, highlighting the diagnostic difficulties that patients with EDC present. Skeletal radiographs were confirmatory in this case and this modality should be emphasized as the simplest and most direct route to the diagnosis. The degree of neurological involvement further distinguishes the case presented from prior reports in the literature. The multiple bilateral intraaxial lesions were intensely enhancing on contrast CT scans, distributed infra- and supratentorially, involving both white and gray matter, and associated with diffuse cerebral edema. The case presented is also remarkable by virtue of the symmetrical involvement of the caudate nuclei, representing the first such example documented in the literature. The diagnosis, treatment, and outcome in this patient are discussed, and a review of the literature is presented.	15352612
2004	14: Endocr Pathol;15(2):15 9-66	Pituitary pathology in Erdheim-Chester disease.	Kovacs K, Bilbao JM, Fornasier VL, Horvath E	Department of Laboratory Medicine and Pathobiology, St. Michael's Hospital, University of Toronto, Toronto, Ontario M5B 1W8, Canada. kovacsk@smh.toronto.on.ca	We report here the histologic and immunohistochemical findings in the autopsy obtained pituitary of a 35-yr-old woman with extensively disseminated Erdheim-Chester disease. It can be concluded that prolactin cell hyperplasia may be the only finding in the adenohypophysis of patients with disseminated Erdheim-Chester disease. It appears that in our patient the clinically apparent anterior hypopituitarism was not due to the lack of storage but rather to insufficient release of adenohypophysial hormones caused by the defect in hypothalamic regulation.	15299202

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2004 Jun	15: Arch Pathol Lab Med;128(6):682-5	Myocardial involvement in Erdheim-Chester disease.	Loeffler AG, Memoli VA	Department of Pathology, Dartmouth-Hitchcock Medical Center, Lebanon, NH 03756, USA. agl@hitchcock.org	While the osseous and systemic changes have been well documented in the current literature, pathologic changes in the myocardium have not been well characterized since Erdheim and Chester's first description of this disease in 1930. In the 2 autopsy cases from Dartmouth-Hitchcock Medical Center (Lebanon, NH) reported in the present study, myocardial involvement was severe and had contributed significantly to the patients' morbidity and death. We describe the autopsy results and correlate them with Erdheim's original descriptions of this disease. In neither of our cases was bony involvement characteristic of the disease, and the diagnosis was made postmortem on the basis of soft tissue findings at autopsy.	15163229
2004 Jun	16: J Neurosurg;100(6):1115-8	Erdheim-Chester disease mimicking a primary brain tumor. Case report.	Rushing EJ, Bouffard JP, Neal CJ, Koeller K, Martin J, Ozdemirli M, Mena H, Ecklund JM	Department of Neuropathology, Armed Forces Institute of Pathology, Washington, DC 20306-6000, USA. rushinge@afip.osd.mil	Case of a 26-year-old man diagnosed with seizures and a well-circumscribed temporoparietal mass that had been demonstrated on imaging studies. Both preoperative and intraoperative diagnoses were consistent with a low-grade astrocytic neoplasm. Subsequent pathological examination indicated a histiocytic proliferation positive for CD68 and factor VIII, and negative for CD1a and S100, with Touton giant cells characteristic of ECD. This case represents the first isolated occurrence of intracranial ECD and its potential to mimic glial neoplasms.	15200134
2004 May	17: Clin Ter;155(5):205-8	[Erdheim-Chester disease: a non-Langerhans cell histiocytosis. A clinical-case and review of the literature]	Valentini D, Cappelli C, Mizzoni F, Noto C, Toscano D, Foco M, Trasimeni G	Servizio di Oncologia Clinica Pediatrica, Università degli Studi di Roma La Sapienza, Roma, Italia. tvjfel@tin.it	We report a case of Erdheim-Chester disease and review 60 cases from the literature. These cases are consider to have Erdheim-Chester disease when they have either typical bone radiographs (symmetrical long bones osteosclerosis) and/or histologic criteria disclosing histiocytic infiltration with distinctive immunohistochemical phenotype of the non-Langerhans cell histiocytes with positive staining for CD68 and negative staining for S-100 protein and CD1a. Our patient undergoes chemiotherapy according to the LCH-II stratification and therapy plan (Vinblastine, Etoposide and Prednisone) and thereafter receives Carboplatin and Etoposide, and Somatostatin. She is alive and clinically well 33 months after onset of symptoms and the lesions don't appear to progress at imaging examinations. In conclusion, Erdheim-Chester disease may be confused with Langerhans cell histiocytosis as it sometimes shares the same clinical (exophthalmos, diabetes insipidus) or radiologic (osteolytic lesions) findings. However, the characteristics radiological pattern of Erdheim-Chester disease together the immunohistochemical phenotype of hystiocytic infiltration supports the theory that Erdheim-Chester disease is a unique disease entity distinct.	15344569
2004 May	18: Rev Neurol (Paris);160(5 Pt 1):585-8	[Cerebral Erdheim-Chester disease]	Taillia H, de Greslan T, Adem C, Talarmin F, Renard JL, Flocard F	Service de Neurologie, Hôpital d'Instruction des Armées du Val-de-Grâce, Paris.	We report the case of a 26-old-year man hospitalized for first partial complex epileptic seizure. Brain MRI showed an asymptomatic pseudo-tumor lesion in the brainstem. Diabetes insipidus, hypophyseal gonadotropic deficiency and osteosclerosis of long bones strongly suggested Erdheim-Chester disease, a rare histiocytosis, confirmed after tibial biopsy. Six months later, the patient remained stable. A persistent, and even increased, enhancement with Gd-DTPA on brain MR images was noted as previously described. The review of the literature collected 64 cases, and only 7 cases of cerebral "tumor".	15269681
2004 Apr	19: AJNR Am J Neuroradiol;25(4):627-30	Erdheim-Chester disease: MR imaging, anatomic, and histopathologic correlation of orbital involvement.	De Abreu MR, Chung CB, Biswal S, Haghighi P, Hesselink J, Resnick D	Department of Radiology, Hospital Mae de Deus e Mae de Deus Center, Porto Alegre, Brazil.	Erdheim-Chester disease (ECD) is a rare form of histiocytosis of unknown origin characterized by tissue infiltration by lipid-laden histiocytes. Typically, the diaphyseal and metaphyseal portions of the tubular bones are affected, leading to a characteristic radiographic pattern of bone sclerosis. Orbital involvement is not infrequent and is manifested by exophthalmos and periorbital xanthomatous lesions, with associated visual problems. This case report documents imaging and pathologic findings in a patient with ECD with extensive orbital involvement.	15090356

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2004 Feb	20: Clin Rheumatol;23(1):52-6	Improvement of Erdheim-Chester disease in two patients by sequential treatment with vinblastine and mycophenolate mofetil.	Jendro MC, Zeidler H, Rosenthal H, Haller H, Schwarz A	Department of Rheumatology, Medical School Hannover, Hannover, Germany. michael.jendro@uniklinik-saarland.de	We report two patients who presented initially with different clinical symptoms. The presenting complaint of the first patient was bone pain, predominantly in the legs, whereas in the other patient the initial symptoms were related to obstruction of both ureters, as in idiopathic retroperitoneal fibrosis. Ultimately, ECD was diagnosed in both patients by the occurrence of both pathognomonic manifestations, the histologic presence of non-Langerhans' histiocytes in bone biopsies, and osteosclerotic lesions of the long bones. Because the extraosseous manifestations progressed and a single application of corticosteroids was ineffective, sequential treatment with vinblastine and mycophenolate mofetil, together with prednisolone, was started. At follow-up respectively 15 and 16 months after the start of treatment a beneficial effect was noted in both patients. These cases illustrate the clinical spectrum of ECD, detail the pathognomonic manifestations of this rare disease, emphasize the need to consider ECD as an uncommon but important differential diagnosis in patients with arthralgias or systemic fibrosis, and give the first evidence for a new treatment option.	14749985
2004 Feb	21: Recenti Prog Med;95(2):104-7	[Erdheim-Chester disease]	Caramaschi P, Biasi D, Lestani M, Carletto A, Bonella F, Bambara LM	Dipartimento di Medicina Clinica e Sperimentale, Università, Verona.	**After the observation of 2 cases we have reviewed the literature; we think useful to present the principal features of the disease, which is likely more frequent than expected; Erdheim-Chester disease is rarely diagnosed because of the poor knowledge of the disease, which is not reported on the common textbooks of medicine.**	15072396
2004 Jan	22: AJNR Am J Neuroradiol;25(1):134-7	Erdheim-chester disease mimicking multiple meningiomas syndrome.	Johnson MD, Aulino JP, Jagasia M, Mawn LA	Department of Pathology, Vanderbilt Medical School, Nashville, TN 37232, USA.	We describe a rare case of non-Langerhans histiocytosis, consistent with Erdheim-Chester disease (ECD), which presented with lesions resembling multiple meningiomas. The patient was initially evaluated for migraine headaches. Initial MR imaging demonstrated a parasellar mass and a second mass near the torcula considered to represent meningiomas. Within 1 year, he developed bilateral orbital lesions surrounding both optic nerves, which were also considered meningiomas. Biopsy of one orbital mass revealed a non-Langerhans histiocytosis. Subsequently, soft tissue masses, a pericardial effusion, and bone lesions consistent with ECD were identified.	14729543
2004	23: Mol Imaging Biol;6(1):63-7	Positron emission tomography/computed tomography of a rare xanthogranulomatous process: Erdheim-Chester disease.	Pereira Neto CC, Roman C, Johnson M, Jagasia M, Martin WH, Delbeke D	Department of Radiology, Vanderbilt University Medical Center, Nashville, TN, USA.	A 37-year-old male with cerebral and periorbital lesions was diagnosed with this rare disease and was evaluated with magnetic resonance imaging (MRI) and 2-deoxy-2-[(18)F]fluoro-D-glucose (FDG) with positron emission tomography/computed tomography (PET/CT) imaging at baseline and following therapy. FDG-PET imaging allowed accurate evaluation of the extent of the disease at baseline, as well as assessment of response to therapy.	15018830

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2004	24: Angiol Sosud Khir;10(4):22-9	A comparative study of the aortic wall in patients with Marfan's syndrome and Erdheim's disease.	Sheremet'eva GF, Ivanova AG, Belov IuV, Gens AP, Kocharian EZ		The materiel of intraoperative biopsies (378) and autopsies was used to study the morphology of the aortic wall in patients with Marfan's syndrome (62) and Erdheim's disease (133). Histological, histochemical and electron microscopy research methods were employed with an assay of the results obtained. It has been demonstrated that in connective tissue dysplasia in the aorta, the histological structure of the intima undergoes substantial changes alongside the pathology of the tunica media. It is marked by the formation of a focal and diffuse thickening with the predominance of the interstitial substance, rich in glycosaminoglycans, and of a small amount of fibrous structures. The morphological signs of the impairment of endothelial permeability (vacuolisation of cytoplasm, thinning of its peripheral portions and basal membrane, expansion of intercellular fissures) are identifiabe, which leads to the disorder of aortic wall metabolism and early development of atherosclerosis. The changes in the intima together with the pathology of the tunica media ere predisposing factors of aneurysm formation, dissection and rupture of the aorta.	15627131
2004 Jun	27: Br J Ophthalmol;88(6):844-7	Treatment of Erdheim-Chester disease with cladribine: a rational approach.	Myra C, Sloper L, Tighe PJ, McIntosh RS, Stevens SE, Gregson RH, Sokal M, Haynes AP, Powell RJ			15148234
2004 Apr	28: J Clin Neurosci;11(3):288, 299	Images in neuroscience: question. Erdheim-Chester disease.	DH Ma	Neurology Department, Royal Melbourne Hospital, Australia.		14975419
2004 Jul	31: Ophthal Plast Reconstr Surg;20(4):329-32	Adult orbital xanthogranuloma with associated adult-onset asthma.	Hammond MD, Niemi EW, Ward TP, Eiseman AS	Ophthalmic Services, Walter Reed Army Medical Center, 6900 Georgia Avenue, Washington, D.C. 20307-5001, U.S.A.	: The authors report a case of adult orbital xanthogranuloma with associated adult-onset asthma in a 44-year-old man. Adult orbital xanthogranuloma was diagnosed on the basis of the clinical findings of bilateral, indurated, yellow eyelid lesions in a patient presenting with adult-onset asthma. Incisional biopsy of the eyelid lesions demonstrated a diffuse histiocytic infiltrate of the orbit and Touton giant cells without evidence of necrobiosis. Systemic evaluation failed to show evidence of bone lesions or paraproteinemia. When patients present with atypical indurated yellow eyelid lesions, a biopsy should be considered. If Touton giant cells are present, a systemic evaluation should be undertaken to rule out both Erdheim-Chester disease and necrobiotic xanthogranuloma. If no systemic findings are present, other than the possibility of adult-onset asthma, the rare entity of adult orbital xanthogranuloma should be considered.	15266154
2003 Dec	2: Clin Rheumatol;22(6):464-6	Aggressive and atypical manifestations of Erdheim-Chester disease.	Lyders EM, Kaushik S, Perez-Berenguer J, Henry DA	Medical College of Virginia Hospital, Virginia Commonwealth University, Richmond, VA 23298-0615, USA.	Erdheim-Chester disease (ECD) is a disseminated non-Langerhans' cell histiocytosis with multisystem involvement, including characteristic sclerotic musculoskeletal lesions. We present the case of a 27-year-old woman with a fulminant course and atypical involvement by ECD manifesting as extensive cerebrovascular disease and lytic musculoskeletal lesions. This case represents an unusual and aggressive presentation of ECD owing to the patient's young age, the severity of the cerebrovascular involvement and the lytic osseous lesions.	14677030

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2003 Nov	3: Thorax;58(11):1004-5	Erdheim-Chester disease: pulmonary infiltration responding to cyclophosphamide and prednisolone.	Bourke SC, Nicholson AG, Gibson GJ	Consultant Respiratory Physician, Northumbria Healthcare Trust and Freeman Hospital, Newcastle upon Tyne, UK. sbourke@doctors.org.uk	We report the case history of a 55 year old man with this condition with extensive and progressive pulmonary disease. He had no response to prednisolone alone, but treatment with prednisolone plus cyclophosphamide was associated with a rapid improvement in symptoms, lung function, and the chest radiographic appearance. He subsequently showed a symptomatic, functional, and radiological deterioration, followed by a marked slowing in the rate of decline. He currently remains stable 41 months after treatment was initiated. This is the first report of pulmonary Erdheim-Chester disease showing improvement in both lung function and symptoms with any form of treatment.	14586060
2003	4: Clin Neuropathol;22(5):246-51	Extracerebral subdural manifestation of Chester-Erdheim disease associated with a giant adenoma of the pituitary.	Thorns V, Zajaczek J, Becker H, Walter GF, Hori A	Institute of Neuropathology, Hannover Medical School, Hannover, Germany. Thorns.veronika@mh-hannover.de	Here we present a case of a 55-year-old woman who developed an exophthalmus, edema and dyspnea, finally leading to death 4 months post admission to the hospital. A cMRI showed a retrobulbar fibrosis, a tumor in the sella turcica, and further tumor formation expanding from the pons to the spinal cord, but without involvement of the dural sheet. Autopsy revealed multiple tumors attached to the pituitary gland, the tentorium, and the brainstem as well as a diffuse thickening of the dura. Histologically, the tumor tissue consisted of densely packed lipid-laden foamy macrophages positive for CD68 and intervening fibrillary cords. Interestingly, tumor cells did not infiltrate/affect the parenchyma but showed a strictly extracerebral/ subdural location. In addition, sections of the pituitary tumor revealed a chromophobe giant adenoma of the pituitary gland. As to our knowledge this is the first detailed description of an exceptional case of intracranial CED presenting with strictly extracerebral/subdural tumor masses accompanied by a giant adenoma of the pituitary gland.	14531550
2003 Aug	5: Arch Pathol Lab Med;127(8):e337-9	Erdheim-Chester disease: a unique presentation with liver involvement and vertebral osteolytic lesions.	Ivan D, Neto A, Lemos L, Gupta A	Department of Pathology and Laboratory Medicine, Medical School, University of Texas, Houston, Tex 77030, USA. doina.ivan@uth.tmc.edu	Although most of the cases have symmetric osteosclerosis of the long bones, an involvement of the axial skeleton has also been described. Extraskeletal lesions are present in more than 50% of the patients and may involve the retroperitoneal space, lungs, kidneys, brain, retro-orbital space, and heart. This study presents the case of a patient with Erdheim-Chester disease with vertebral destruction and, for the first time, to our knowledge, involvement of the liver. The diagnosis is based on radiologic, histologic, immunohistochemical, and ultrastructural findings.	12873197
2003 Aug	6: Australas J Dermatol;44(3):194-8	Erdheim-Chester disease.	Opie KM, Kaye J, Vinciullo C	Department of Dermatology, Royal Perth Hospital, Perth, Western Australia, Australia.	A 38-year-old man presented with numerous dermal nodules, similar to xanthoma disseminatum, that were histologically consistent with his diagnosis of Erdheim-Chester disease, a non-Langerhans cell histiocytosis. Other cutaneous manifestations of this disease include eyelid xanthelasma, pretibial dermopathy and pigmented lesions of the lips and buccal mucosa. The histological diagnosis of Erdheim-Chester disease was originally made on the patient's retroperitoneal tissue, obtained at a laparotomy for surgical treatment of a presumed phaeochromocytoma, and confirmed by the pathognomonic long bone X-ray findings of this disease.	12869045
2003	7: Rev Esp Med Nucl;22(4):253-6	[Bone scintigraphy in Erdheim-Chester disease]	Pena Pardo FJ, Banzo Marraco I, Quirce Pisano R, Hernández Allende R, Carril Carril JM	Servicio de Medicina Nuclear. Hospital Universitario Marqués de Valdecilla. Universidad de Cantabria. Santander. España. mnuccj@humv.es	In this work, we report 2 ECD cases and their respective bone scans showing typical findings described in the literature. We found bilateral and symmetrical increased uptake of diaphyses and metaphyses of long bones, mainly in lower limbs. The mid-diaphyses and the epiphyses (partially in the first case) as well as the axial skeleton are spared.	12846951

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2003 Apr	8: Neuroradiology; 45(4):241-5	Cerebral Erdheim-Chester disease: case report and review of the literature.	Weidauer S, von Stuckrad-Barre S, Dettmann E, Zanella FE, Lanfermann H	Institute of Neuroradiology, University of Frankfurt, Schleusenweg 2-1660528, Frankfurt, Germany. Weidauer@em.uni-frankfurt.de	We report on a 44-year-old man with biopsy-proven Erdheim-Chester disease and slowly progressive cerebellar dysfunction. MRI showed additional symmetrical hyperintense signal changes in the superior cerebellar peduncle as well as in the trigonum lemnisci on coronal FLAIR images. The widespread neurological manifestations of cerebral Erdheim-Chester disease and differential diagnosis are discussed.	12687308
2003	9: Clin Exp Rheumatol;21(2):232-6	Biochemical markers of bone turnover, serum levels of interleukin-6/interleukin-6 soluble receptor and bisphosphonate treatment in Erdheim-Chester disease.	Mossetti G, Rendina D, Numis FG, Somma P, Postiglione L, Nunziata V	Department of Clinical and Experimental Medicine, Federico II University Medical School, Naples, Italy. nunziata@unina.it	Erdheim-Chester disease (ECD) is a rare non-Langherans form of histiocytosis characterized radiologically by symmetrical sclerosis of the metaphysis and the diaphysis of long tubular bones. Macrophages are potent interleukin-6 (IL-6) producers and elevated IL-6 serum levels have been described in pathological conditions characterized by increased bone resorption. In a patient with ECD, during the acute phase of the disease we found high serum levels of IL-6 and IL-6 soluble receptor (sIL-6R) and high levels of bone turnover markers. After 5 years of combination therapy with oral prednisone and intravenous clodronate a significant reduction in the above mentioned biological parameters was seen. We suggest that the systemic disorders present in ECD could be related to the high serum levels of IL-6 and sIL-6R. We also propose the use of bisphosphonates in the clinical management of ECD.	12747282
2003	10: J Cutan Med Surg;7(2):129-32	Erdheim-Chester disease.	Lenahan SE, Helm KF, Hopper KD	Department of Pathology, The Milton S. Hershey Medical Center, Penn State University, Hershey, Pennsylvania, USA.	BACKGROUND: Erdheim-Chester disease is a rare non-Langerhans' cell histiocytosis. OBJECTIVE: This case report is presented to familiarize clinicians with Erdheim-Chester disease and its differential diagnosis. RESULTS AND CONCLUSION: Erdheim-Chester disease presents with unique clinical and pathologic findings. Its xanthoma-like lesions can cause significant morbidity and mortality.	12447617
2003 Feb	11: Eur J Intern Med;14(1):53-55	Erdheim-Chester disease: a rare cause of paraplegia.	Curgunlu A, Karter Y, Oztürk A	Department of Internal Medicine, Istanbul University Cerrahpaşa Medical Faculty, Istanbul, Turkey	Until now, only two cases of Erdheim-Chester disease with paraparesis have been reported. Herein we report the first case of Erdheim-Chester disease with the clinical manifestation of paraplegia. Our patient also had diabetes insipidus, pleural and pericardial effusion, retro-orbital and cavernous sinus masses, fibrotic changes in the retroperitoneal, perirenal, and periaortic areas, and epidural space-occupying lesions. We want to emphasize that ECD may be a very rare cause of paraplegia.	12554012
2003 Dec	12: Med Pediatr Oncol;41(6):575-7	Erdheim-Chester disease in a child.	Clerico A, Ragni G, Cappelli C, Schiavetti A, Gonfiantini M, Uccini S	Oncology Service, Pediatric Clinic, University of Rome "La Sapienza", Rome, Italy.		14595723
2003 Jul	13: Rofo;175(7):992-3	[Erdheim-Chester disease: radiologic diagnosis]	Niehues SM, Riechert FC, Lemke AJ			12847657
2003 May	14: Adv Anat Pathol;10(3):160-71	Erdheim-Chester disease: clinical and pathologic spectrum of four cases from the Arkadi M. Rywlin slide seminars.	Bisceglia M, Cammisa M, Suster S, Colby TV	Servizio di Anatomia Patologica and dagger Dipartimento di Scienze Radiologiche, IRCCS-Ospedale Casa Sollievo della Sofferenza, San Giovanni Rotondo (FG), Italy. bismi@libero.it		12717118

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2003 May	15: Orthopedics;26(5):505-8	Erdheim-Chester disease.	Papagelopoulos PJ, Savvidou OD, Galanis EC, Wenger DE, Inwards CY, Sim FH	First Department of Orthopedic Surgery, Athens University Medical School, Athens, Greece.		12755215
2003 Apr	16: J Urol;169(4):1470-1	Erdheim-Chester disease: case report and review of associated urological, radiological and histological features.	Yun EJ, Yeh BM, Yabes AP, Coakley FV, Kane CJ	Deaprtment of Urology, University of California, San Francisco Medical School, USA.		12629387
2003 Mar	17: Ann Rheum Dis;62(3):271-2	Erdheim-Chester disease with early onset atherosclerosis and a pseudo-malignant phase.	Neame RL, Struthers GR			12594120
2003 Mar	18: Ann Rheum Dis;62(3):270	Erdheim-Chester disease in Brazil.	Lopes Marques CD, Duarte AL, Cavalcanti Fd Fde S			12594119
2003 Sep	19: Ophthal Plast Reconstr Surg;19(5):372-81	Orbital xanthogranuloma: clinical and morphologic features in eight patients.	Karcioglu ZA, Sharara N, Boles TL, Nasr AM	Tulane University Health Sciences Center, Departments of Ophthalmology and Pathology, New Orleans, Louisiana, USA.	PURPOSE: To describe the clinical and morphologic features of patients with orbital xanthogranuloma (XG) with or without Erdheim-Chester disease (E-Cd). Two patients with E-Cd with involvement of the long bones of the upper and lower extremities and retroperitoneal region died of kidney failure within approximately 1 year of diagnosis. CONCLUSIONS: Orbital XG is a proliferative lesion of the non-Langerhans histiocytes, which may present as a solitary orbital lesion or may be associated with a systemic condition known as E-Cd with very poor prognosis.	14506422
2003 Apr	21: J Thorac Imaging;18(2):116-21	Notes from the 2002 annual meeting of the Korean Society of Thoracic Radiology.	Kang EY, Choi YH, Im JG, Park CK	Department of Diagnostic Radiology, Korea University Guro Hospital, Seoul, Korea. keyrad@korea.ac.kr		12700490
2002 Dec	2: Neuroradiology;44(12):1004-7	Spinal dural involvement in Erdheim-Chester disease: MRI findings.	Albayram S, Kizilkilic O, Zulfikar Z, Islak C, Kocer N	Department of Radiology, Division of Neuroradiology, The Cerrahpasa Medical School, 34300 Kocamustafapasa, Istanbul, Turkey. salbayram@hotmail.com	There are very few reported cases of Erdheim-Chester disease that document involvement of dura at the level of the spinal cord. Among these reports, we know of no publication that includes detailed MRI findings. To the best of our knowledge, the case presented here is the first published report of this specific manifestation of Erdheim-Chester disease that includes detailed MRI findings in addition to the related history. Spinal manifestations of Erdheim-Chester disease in our patient were at the dorsal and lumbar levels (T1-T6 and T12-T11 respectively). Both epidural and subdural linear large masses were present, causing spinal cord compression at the dorsal level and epidural thickening at the lumbar level.	12483447

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2002 Sep	3: J Endocrinol Invest;25(8):727-9	Erdheim-Chester syndrome, presenting as hypogonadotropic hypogonadism and diabetes insipidus.	Khamseh ME, Mollanai S, Hashemi F, Rezaizadeh A, Azizi F	Endocrine Research Center, Shaheed Beheshti University of Medical Sciences, Tehran, IR Iran.	It is still a matter of discussion whether Erdheim-Chester syndrome is a distinct entity or a type of LCH. The present case is a 46-yr-old man, that presented with signs and symptoms of diabetes insipidus and hypogonadotropic hypogonadism simultaneously. X-rays and bone scintigraphy showed typical and pathognomic findings of Erdheim-Chester syndrome. Bone biopsy and immunohistochemical staining strongly support the diagnosis of non-Langerhans cell histiocytosis.	12240906
2002 Sep	4: J Neurooncol;59(2):169-72	Failure of radiation therapy for brain involvement in Erdheim Chester disease.	Mascalchi M, Nencini P, Nistri M, Sarti C, Santoni R	Dipartimento di Fisiopatologia Clinica, Università di Firenze, Italia. m.mascalchi@dfc.unifi.it	A patient with suprasellar and brain stem involvement in Erdheim Chester disease (ECD) underwent magnetic resonance (MR) imaging and proton MR spectroscopy (1H MRS) of the ventral pons before and 1, 4 and 18 months after external whole-brain (24 Gy) radiotherapy. By revealing a decrease of the N-acetyl-aspartate/choline ratio in the pons, 1H MR spectroscopy anticipated lesions growth on MR imaging. In line with the results in four patients reported in the literature, our observation indicates that external radiation therapy is not effective for intracranial involvement in ECD.	12241111
2002 Aug	5: Am J Med Sci;324(2):96-100	Erdheim-Chester disease with prominent pericardial involvement: clinical, radiologic, and histologic findings.	Gupta A, Kelly B, McGuigan JE	Department of Medicine, University of Florida College of Medicine, Gainesville 32610, USA.	We describe documented skeletal and pericardial involvement by ECD producing cardiac tamponade in a 30-year-old woman. The patient presented with jaundice and hepatic congestion produced by cardiac tamponade. Pericardial biopsy revealed xanthogranulomatous lesions comprised of foamy and lipid-laden macrophages, multinucleated giant cells, monocytes, and lymphocytes in a mesh of fibrosis. Immunohistochemical staining was positive for CD68 and negative for CD1a, consistent with ECD rather than with the much more common Langerhans cell form of histiocytosis.	12186113
2002 Jun	6: Mod Pathol;15(6):666-72	Erdheim-Chester disease: case report, PCR-based analysis of clonality, and review of literature.	Al-Quran S, Reith J, Bradley J, Rimsza L	Department of Pathology, Immunology and Laboratory Medicine, University of Florida College of Medicine, Shands Hospital, Gainesville 32610-0275, USA.	We present a case report of ECD in a 35-year-old African-American woman with a progressive course over 6 years. We investigated the clonality of the histiocytes using the HUMARA assay on paraffin-embedded tissue sections but did not find any evidence that these cells represent a monoclonal population. In this report, the characteristics of ECD are reviewed, the genetic basis of the HUMARA assay is discussed, and our results in the context of other clonality investigations reported in the literature to date are summarized.	12065781
2002	7: J Comput Assist Tomogr;26(2):257-61	MR findings of Erdheim-Chester disease.	Gottlieb R, Chen A	Westlake Diagnostic Center, Thousand Oaks, CA, USA. roymay@aol.com	Lipoid granulomatosis (Erdheim-Chester disease) is a rare but distinct form of histiocytosis. This disease has characteristic radiologic findings involving the musculoskeletal system that are critical to the diagnosis: symmetric sclerosis of the metaphysis and diaphysis of long bones with relative sparing of the epiphysis as depicted on conventional radiography. However, it is a systemic disease that involves multiple organ systems. This pictorial essay is of a single patient imaged over multiple years, using various pulse sequences with both low and high field strength MR scanners. It depicts many of the characteristic findings encountered in this rare systemic disorder.	11884783
2002 Feb	8: J Neurosurg;96(2):344-51	Multiple system Erdheim-Chester disease with massive hypothalamic-sellar involvement and hypopituitarism.	Oweity T, Scheithauer BW, Ching HS, Lei C, Wong KP	Department of Pathology, Normah Medical Specialist Center, Kuching, Sarawak, Malaysia.	The authors report a case of ECD in which the diagnosis was made after biopsy of a hypothalamic mass. The mass had been discovered during a workup for panhypopituitarism in a 55-year-old man with urological and bone disease. Aside from diabetes insipidus, other features of pituitary insufficiency have seldom been reported and no patients have presented with a hypothalamic tumor. The endocrinological and neurological aspects of ECD are discussed, as is its differential diagnosis. Reported cases of the disorder associated with hypopituitarism or found during biopsy of central nervous system structures are also reviewed.	11838810

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2002	9: Clin Neuropathol;21(1):24-8	Xanthogranuloma of the Erdheim-Chester type within the sellar region: case report.	Reithmeier T, Trost HA, Wolf S, Stölzle A, Feiden W, Lumenta CB	Department of Neurosurgery, Academic Hospital Bogenhausen, Technical University of Munich, Germany. Thomas.Reithmeier@web.de	Manifestations of Erdheim-Chester disease in the central nervous system are very rare. Cases with localization in the retroorbital space, hypothalamic area and posterior pituitary as well as intracerebral lesions are known. In our neurosurgical unit, a 51-year-old male patient with a history of hypophyseal insufficiency and visual deficits underwent surgery for a pituitary lesion. Histological and immunohistochemical examination revealed a xanthogranulomatous lesion composed of very large CD68-positive foam cells with small nuclei and some Touton-like giant cells, histiocytes, as well as loci with small lymphocytes and isolated eosinophilic granuolcytes, embedded in fibrotic tissue. Based on these findings, the histological diagnosis was a xanthogranuloma of the Erdheim-Chester type.	11846041
2002 Jan	10: J Fr Ophtalmol;25(1):57-61	[Bilateral exophthalmos diabetes insipidus: Erdheim-Chester disease. Clinical and radiological findings]	Le Goff L, Berros P, Denis D, Ridings B	Service d'Ophtalmologie de Marseille, CHU Timone, 264, rue Saint-Pierre, 13385 Marseille, France.	The authors report a case of a 61-year-old man presenting bilateral exophthalmos and diabetes insipidus. A retro-orbital biopsy revealed nonspecific fibrocollagenic infiltration. The diagnosis of Erdheim-Chester disease was evoked when a multivisceral affection (retroperitoneal and mediastinal periaortic fibrosis) with specific bone localization became evident. The histopatholgical study of a bone biopsy showed xanthogranulomatous infiltration. The patient died a few months later of an intercurrent infection.	11965120
2002 Mar	11: Ann Rheum Dis;61(3):199-200	Erdheim-Chester disease: typical radiological bone features for a rare xanthogranulomatosis.	Breuil V, Brocq O, Pellegrino C, Grimaud A, Euller-Ziegler L	Rheumatology Department, l'Archet University, 06200 Nice, France.		11830422
2002 Mar	12: J Bone Miner Res;17(3):381-3	Imaging of Erdheim-Chester disease.	Olmos JM, Canga A, Velero C, González-Macías J	Department de Medicina Interna, Hospital Marqués de Valdecilla, Universidad de Cantabria, Santander, Spain.		11874230
2002 Feb	13: AJR Am J Roentgenol;178(2):429-32	Erdheim-Chester disease: a unique presentation with multiple osteolytic lesions of the spine and pelvis that spared the appendicular skeleton.	Klieger MR, Schultz E, Elkowitz DE, Arlen M, Hajdu SI	Department of Radiology, North Shore University Hospital, 300 Community Dr., Manhasset, NY 11030, USA.		11804910
2002 Aug	14: Exp Clin Endocrinol Diabetes;110(5):248-52	Psychoneuroendocrine disturbances in a patient with a rare granulomatous disease.	Perras B, Petersen D, Lorch H, Fehm HL	Department of Internal Medicine I, Universität zu Lübeck, Ratzeburger Allee, Germany. Perras@kfg.mu-luebeck.de	We report on a patient with the clinical diagnosis of ECD displaying endocrine and cerebral manifestations and skeletal, pulmonary and soft tissue involvement. Disturbance of the endocrine system was revealed by enlargement of the pituitary, partial deficiency of growth hormone (GH), hyperprolactinemia and testosterone deficiency. Cerebral involvement included sinus vein thrombosis, pathologic acoustic evoked potentials, persistence of gadolinium enhancement after magnetic resonance imaging and hypomania. These findings emphasize the importance to assess endocrine and cerebral function in patients with rare granulomatous diseases like ECD and multiorgan involvement.	12148090

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2001 Dec	1: Am J Ophthalmol;132 (6):945-7	Interferon therapy for orbital infiltration secondary to Erdheim-Chester disease.	Esmaeli B, Ahmadi A, Tang R, Schiffman J, Kurzrock R	Ophthalmology Section, Department of Plastic Surgery, The University of Texas M.D. Anderson Cancer Center, 1515 Holcombe Blvd., Houston, TX 77030, USA. besmaeli@mdanderson .org	PURPOSE: To describe a 55-year-old male with Erdheim-Chester disease with bilateral orbital infiltration and visual loss who was successfully treated with interferon-alpha. METHODS: Interventional case report. RESULTS: The patient was treated with interferon-alpha and had an improvement in his clinical signs, including his visual acuity, after 4 weeks of interferon therapy. CONCLUSION: Interferon-alpha can be effective in the treatment of orbital infiltration secondary to Erdheim-Chester disease.	11730673
2001 Dec	2: Ann Pathol;21(6):52 9-33	[Erdheim-Chester disease. Apropos of a case with autopsy findings]	Ranty ML, Le Pessot F, Billerey C, Dominique S, Métayer J	Laboratoire d'Anatomie et de Cytologie Pathologiques, CHU Charles Nicolle, Boulevard Gambetta, 76031 Rouen.	Erdheim-Chester's Disease is a very uncommon variety of non-Langerhans histiocytosis of unknown etiology, which characteristically affects long bones bilaterally and symmetrically in adults. It may be accompanied by visceral foci of variable localization and extension determining prognosis. Bone scintigraphy is characteristic enough to evoke the disease but histologic examination of a peripheral specimen is required to confirm the diagnosis: spumous histiocytes CD68+, PS100+/-, CD1a-. We describe a case revealed by a severe lung disease with detailed autopsy.	11910940
2001 Sep	3: Yan Ke Xue Bao;17(3):163-7	A case of Erdheim-Chester disease with bilateral orbital involvement.	Wu Z, Yan J, Hong W, Yuan Y, Dai L	Zhongshan Ophthalmic Center, Sun Yat-sen University of Medical Sciences, Guangzhou, 510060 China.	A 43-year-old female with bilateral proptosis was presented. CT demonstrated bilateral, diffuse orbital mass. Histopathologic assessment revealed a diffuse xanthogranulomatous process with clusters of lipidladen histocytes. Numerous Touton giant cells were scattered throughout the lesion. Renal and heart failure happened during a 6-year follow-up period. Long bones roentgenogram demonstrated diffuse symmetrical sclerosis with extensive, lytic lesions. Systemic administration of corticosteroids, chemotherapy, immunoglobulin and traditional Chinese medicine showed good therapeutic result. CONCLUSIONS: An administration of systemic corticosteroids, chemotherapy, immunoglobin and traditional Chinese medicine can control Erdheim-Chester disease. Further exploration of its pathogenesis and collection of useful clinical data are required.	12567744
2001	4: Am J Nephrol;21(4):3 15-7	Two enlarged kidneys: a manifestation of Erdheim-Chester disease.	André M, Delèvaux I, de Fraissinette B, Ponsonnaille J, Costes Chalret N, Wechsler B, Piette JC, Aumaître O	Department of Internal Medicine, Groupe Hospitalier Saint-Jacques, Clermont-Ferrand, France.	We describe the case of a patient with a pleural and pericardial effusion leading to tamponade. Pathological examination of pericardium and mediastinal adenopathy was normal. The abdominal computed tomography scan showed two enlarged kidneys suggestive of Erdheim-Chester disease. Bone scan scintigraphy demonstrated symmetrical increased labeling of the long bones. The biopsy of perirenal soft tissue confirmed the diagnosis of Erdheim-Chester disease.	11509804
2001 Jul	5: Cesk Patol;37(3):114-7	[Severe pulmonary involvement in Erdheim-Chester disease (case report)]	Z Kinkor	Oddělení patologie Fakultní nemocnice Na Bulovce, Praha.	Presented is a typical case of Erdheim-Chester disease (ECD) wherein the severe pulmonary manifestation led to an open lung biopsy and eventual morphologic recognition of the nonspecific clinical symptoms. The pulmonary involvement is described in almost 20% of cases and is prognostically unfavourable. About 20 cases of Erdheim-Chester disease were published and more than half of them had lethal outcome. The ECD affecting seriously lungs appears as non-specific interstitial lung disease and usually does not enter the broad clinical differential diagnosis. A detailed bibliography with special attention to the pulmonary involvement by this enigmatic disease is presented.	11669020

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2001 Jun	6: Clin Radiol;56(6):481-4	Erdheim-chester disease.	Murray D, Marshall M, England E, Mander J, Chakera TM	Department of Diagnostic Radiology, Royal Perth Hospital, Perth, WA, Australia.	We describe two confirmed cases of ECD, both of which demonstrate non-malignant retroperitoneal and peri-renal infiltration causing dilatation of the upper renal tracts. The cases are illustrated with contrast studies, computed tomography (CT) and magnetic resonance imaging (MRI). Typical sclerosis of the long bones was apparent on radiography. Both cases have been treated conservatively to date.A brief review of the literature regarding the manifestations of ECD is included. In cases of non-malignant retroperitoneal infiltration, ECD should be considered as a diagnosis and radiographs of the long bones performed.	11428798
2001 Jun	7: Hautarzt;52(6):510-7	[Skin manifestations of Erdheim-Chester disease. Case report and review of the literature]	Watermann DF, Kiesewetter F, Frosch PJ	Hautklinik der Städtischen Kliniken Dortmund und Lehrstuhl für Dermatologie der Universität Witten/Herdecke.	A 46 year old woman suffering from Erdheim-Chester disease is Histologically the skin manifestations were also a sign of the basic disease which had spread to various organsFurther localizations of Erdheim-Chester disease were found in the femurs, tibiae and mandibula as well as in the right breast, retroorbital region and abdominal aorta. Infiltration of the retroperitoneal cavity led to urinary retention and nephrectomy. With systemic corticosteroid therapy, the skin lesions on the flanks regressed but recurred after discontinuance of the drug.	11428080
2001 Jun	8: Nervenarzt;72(6):449-52	[Cerebellar syndrome, exophthalmos and secondary hypogonadism in Erdheim-Chester disease]	Grothe C, Urbach H, Bös M, Ko Y, Schröder R	Neurologische Universitätsklinik Bonn, Sigmund Freud Strasse 25, 53105 Bonn. c.grothe@uni-bonn.de	We present a 50-year-old patient with a slowly progressive cerebellar syndrome, left-sided exophthalmos, secondary hypogonadism, and multiple pleomorphous skin alterations. The diagnosis of Erdheim-Chester disease was established by the radiological detection of a left-sided retrobulbar space-occupying mass, a hypophysial stalk lesion, alterations in both cerebellar hemispheres, retroperitoneal imbibition, osteolytic/osteosclerotic changes in the metaphysis and diaphysis of the long bones, and a skin biopsy with histological detection of a non-Langerhans-cell histiocytosis.	11433705
2001 May	9: J Radiol;82(5):580-2	[Retroperitoneal complications of Erdheim-Chester disease]	Leluc O, André M, Marciano S, Lafforgue P, Rossi D, Bartoli J	Service de radiologie, Hôpital Salvator, 270, boulevard Sainte Marguerite, 13009 Marseille.	Retroperitoneal involvement manifests as a mass associated with fibrosis, which is well visualized on CT scan and MRI. This disease is characterized by its potential to involve the whole retroperitoneum. We report a case of this disease that developed over twenty years, consisting of renal arteries stenosis, bilateral ureteral stenosis and evolutive adhesive capsulitis.	11416797
2001 Apr	10: Monaldi Arch Chest Dis;56(2):115-7	Erdheim-Chester disease. A case report.	Vasáková M, Fiala P, Kinkor Z	Institute of Tuberculosis and Respiratory Diseases, Thomayer Faculty Hospital, Prague, Czech Republic. tichadohoda@volny.cz	A 63-year old man had a history of diabetes insipidus, arthralgias and myalgias, weight loss, relapsing fever and malaise. Increased uptake of Tc-99m was found predominantly in distal antebrachia, in distal femurs and in both trochanters and tibias on the bone scintigraphy. The chest radiograph showed reticulonodular pattern and the high resolution computed tomography (HRCT) scans revealed diffuse infiltrative lung disease with small multiple nodules and widening of interlobular septs. Videothoracoscopic lung biopsy and biopsy of tibial lesion were perfomed. The histopathologic examination proved non-Langerhans cell histiocytosis-Erdheim-Chester disease. Treatment with prednisone reduced the pain and fever and improved the vital capacity of the lungs while the changes in the lungs and bones remained unchanged.	11499297

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2001 Jan	11: Am J Med Sci;321(1):66-75	Erdheim-Chester disease: a rare multisystem histiocytic disorder associated with interstitial lung disease.	Shamburek RD, Brewer HB, Gochuico BR	Molecular Disease Branch, National Heart, Lung, and Blood Institute, National Institutes of Health, Bethesda, Maryland 20892-1666, USA. bob@mdb.nhlbi.nih.gov	Erdheim-Chester disease (ECD) is a rare multisystem histiocytosis syndrome of unknown cause that usually affects adults. Histiocytic infiltration of multiple end organs produces bone pain, xanthelasma and xanthoma, exophthalmos, diabetes insipidus, and interstitial lung disease. Differential diagnosis includes Langerhans cell histiocytosis, metabolic disorders, malignancy, and sarcoidosis. ECD can be diagnosed using a combination of clinical and histopathologic findings. Sites of involvement include lung, bone, skin, retroorbital tissue, central nervous system, pituitary gland, retroperitoneum, and pericardium. Symmetrical long bone pain with associated osteosclerotic lesions, xanthomas around the eyelids, exophthalmos, and/or diabetes insipidus suggest ECD. Approximately 35% of patients have associated lung involvement, characterized by interstitial accumulations of histiocytic cells and fibrosis in a predominantly perilymphangitic and subpleural pattern. This pattern distinguishes ECD from other histiocytic disorders involving the lung. The diagnosis is confirmed by tissue biopsies that contain histiocytes with non-Langerhans cell features. In general, the clinical course of patients with this disease varies, and the prognosis can be poor despite treatment. Clinical trials for treatment of ECD have not been conducted and treatment is based on anecdotal experience.	11202482
2001 Jan	12: Int J Surg Pathol;9(1):73-9	Erdheim-Chester disease with extensive marrow necrosis: a case report and literature review.	Kim NR, Ko YH, Choe YH, Lee HG, Huh B, Ahn GH	Department of Diagnostic Pathology, Sungkyunkwan University School of Medicine, Samsung Medical Center, Seoul, Korea.	We report a case of Erdheim-Chester disease with diffuse necrosis leading to difficulty in making a prompt diagnosis. Radiologically, osteosclerotic lesions with osteolytic element involved metadiaphyses of both proximal tibia, and retroperitoneal infiltrations encasing both kidneys, both adrenals, and aorta were found. A biopsy of the tibia showed diffuse infiltration of foamy histiocytes, Touton-type giant cells, and fibroblastic cells associated with extensive coagulative necrosis. Immunohistochemically, foamy histiocytes were positive for CD68 and peanut agglutinin and negative for S-100 protein. A few Langerhans' cells, which were difficult to identify in hematoxylin-eosin stain, were highlighted by immunostain for S-100 protein. The patient received supportive therapy and was alive 1 1/2 years after diagnosis, with newly developed bilateral retrobulbar lesions and worsened heart failure.	11469352
2001	13: Rontgenpraxis; 54(4):148-51	[Involvement of the facial skull in Erdheim-Chester disease]	Kirchner TH, Seipelt G, Vogl TJ	Institut für Diagnostische und Interventionelle Radiologie, Johann Wolfgang Goethe-Universität Frankfurt a. M.	We report on a patient suffering from Erdheim-Chester-disease (ECD). ECD represents a very rare entity with lipogranulomatosis of mesenchymal origin. The most common radiological manifestation is the involvement of the long bones of the extremities. Here we find sclerosis of the spongiosa combined with a thinning of cortical structures. This often results in a small crack of hyperlucency between corticals and spongiosa. Our case demonstrates an involvement of the craniofacial part of the skull showing sclerosis of the upper jaw bone. This manifestation has not yet been reported in the literature.	11883118
2001 Dec	14: Am J Med;111(8):672-3	Cardiac tumor due to Erdheim-Chester disease.	Ammann P, Bösch B, Buchholz S, Genoni M, Laube I, Naegeli B			11755517

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2001 Nov	15: Ann Intern Med;135(9):844-5	Treatment of refractory Erdheim-Chester disease with double autologous hematopoietic stem-cell transplantation.	Boissel N, Wechsler B, Leblond V			11694122
2001	16: Radiol Med (Torino);102(1-2):91-3	[A case of Chester-Erdheim disease with unusual bone involvement]	Dimonte M, Minonne A	Servizio di Radiologia, A.O. Cardinale Giovanni Panico, Tricase, Lecce, Italy.		11677449
2001 Jun	17: J Assoc Physicians India;49:671-3	Erdheim-Chester disease.	Sood A, Jain R, Kumar R, Malhotra R, Chopra P	Department of Endocrinology and Metabolism, All India Institute of Medical Sciences, New Delhi.		11584952
2001 May	18: AJR Am J Roentgenol;176(5):1330-1	Erdheim-Chester disease of the retroperitoneum: a rare cause of ureteral obstruction.	Fortman BJ, Beall DP	The Johns Hopkins Hospital, Baltimore, MD 21286, USA.		11312207
2001 Oct	19: Br J Ophthalmol;85(10):1220-4	A role for methotrexate in the management of non-infectious orbital inflammatory disease.	Smith JR, Rosenbaum JT	Casey Eye Institute, Oregon Health Sciences University, Portland, Oregon 97201-4197, USA. smithjus@ohsu.edu	AIM: To evaluate the clinical usefulness of methotrexate for patients with non-infectious orbital inflammatory disease who fail to respond to systemic corticosteroids and/or orbital irradiation. METHODS: The medical records of patients with non-infectious orbital inflammatory disease who were treated with methotrexate at Oregon Health Sciences University were examined. Methotrexate was administered at a median maximum dose of 20 mg per week (range 15-25 mg per week) in conjunction with folate supplementation. The study cohort included 14 patients (24 eyes) with diagnoses including non-specific orbital inflammation (n=7), Tolosa-Hunt syndrome (n=1), thyroid orbitopathy (n=3), Wegener's granulomatosis (n=1), sarcoidosis (n=1), and Erdheim-Chester disease (n=1). In all cases, methotrexate was commenced as a corticosteroid sparing agent. 10 patients (71%) completed a 4 month therapeutic trial of methotrexate. Median duration of treatment for the nine (64%) patients who experienced clinical benefit was 25 months (range 10-47 months). Six responders were ultimately able to cease methotrexate, including the single patient who required concurrent long term corticosteroid therapy. Complications included fatigue, gastrointestinal disturbance, hair thinning and mild, reversible serum liver enzyme elevation. Two patients (14%) discontinued treatment because of adverse effects. CONCLUSION: Methotrexate is a well tolerated immunosuppressive medication which may benefit patients with recalcitrant non-infectious orbital inflammatory disease.	11567968

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2001 Oct	20: J Clin Endocrinol Metab;86(10):4603-10	Extensive inflammatory pseudotumor of the pituitary.	Hansen I, Petrossians P, Thiry A, Flandroy P, Gaillard RC, Kovacs K, Claes F, Stevenaert A, Piguet P, Beckers A	Department of Neurology, University of Liege, B 4000 Liege, Belgium.	A 40-yr-old female presented with an extensive lesion of the sellar area and the sphenoid sinus, spreading to the optic nerves and associated with pachymeningitis. Histological findings were consistent with an inflammatory pseudotumor, and steroid treatment allowed the disappearance of all the lesions. Inflammatory pseudotumors of the pituitary are very rare. This case appears unique with regard to the extension of the lesions and the dramatic response to medical treatment. The differential diagnosis of inflammatory lesions of the pituitary is difficult. It relies mainly on histological analysis and includes sarcoidosis, Wegener's granulomatosis, histiocytosis (Langerhans, Rosai-Dorfman, and Erdheim-Chester diseases) and lymphocytic hypophysitis.	11600510
2000 Sep	1: Oral Surg Oral Med Oral Pathol Oral Radiol Endod;90(3):389-98	Erdheim-Chester disease of the jaws: literature review and case report.	Petrikowski CG, McGaw WT	Faculty of Dentistry, University of Toronto, Ontario, Canada. grace.petrikowski@utoronto.ca	Erdheim-Chester disease is a rare systemic lipogranulomatous disorder of adults that shares some histopathologic features similar to Langerhans' cell histiocytosis and that results in characteristic radiographic changes in the long bones. Relatively few cases have been reported in the jaws. We present a literature review of jaw cases and the first case report to describe detailed radiographic and pathologic features of jaw involvement, as well as clinical, radiographic, and histopathologic follow-up of the untreated jaw lesions.	10982964
2000 Aug	2: Eye;14 (Pt 4):606-12	Erdheim-Chester disease: two cases of orbital involvement.	Sheidow TG, Nicolle DA, Heathcote JG	Department of Ophthalmology, University of Western Ontario, London, Canada. tgsheido@julian.uwo.ca	We describe two patients, one presenting with diabetes insipidus and subsequently developing orbital pseudotumours and retroperitoneal fibrosis, the other presenting with exophthalmos and diplopia. The first patient was treated with cladribine and subsequently developed sudden onset of bilateral blindness while the second required radiation therapy for the retro-orbital process and developed radiation retinopathy. These cases typify the variable presentation and course in patients with ECD.	11040908
2000 Jul	3: Mod Pathol;13(7):747-54	Pulmonary pathology of Erdheim-Chester disease.	Rush WL, Andriko JA, Galateau-Salle F, Brambilla E, Brambilla C, Ziany-bey I, Rosado-de-Christenson ML, Travis WD	Department of Dermatopathology, Armed Forces Institute of Pathology, Washington, DC 20306-6000, USA.	The clinical, radiologic, and pathologic features of six patients with ECD with lung involvement are presented. The patients were three men and three women (mean age, 57). Five presented with progressive dyspnea, and one presented with diabetes insipidus. Open-lung biopsies showed histiocytic infiltrates in a lymphangitic pattern with associated fibrosis and lymphoplasmacytic inflammatory infiltrates. Clinical follow-up of up to 16 years was available. At the end of that time, five patients were dead of complications related to their disease; one patient remains alive 4 years after diagnosis but with severe respiratory compromise.	10912934
2000 Jun	4: Clin Nucl Med;25(6):414-20	The role of bone scintigraphy in patients with Erdheim-Chester disease.	Gotthardt M, Welcke U, Brandt D, Tontsch D, Barth PJ, Schaefer J, Hoeffken H, Joseph K	Department of Clinical Nuclear Medicine, Philipps-University of Marburg, Germany. gotthard@mailer.uni-marburg.de	Erdheim-Chester disease (ECD) is a rare disorder that has been reported fewer than 60 times in the literature. Although clinical findings seem to be specific at first sight, histologic classification remains unclear. It has not been decided whether ECD is part of the spectrum of histiocytoses or whether it may be a lipid storage disorder or even a primary macrophage cell disorder, although it does show a distinct histologic pattern. However, the clinical appearance alone shows several typical features, rendering the diagnosis very probable if present. This article illustrates the importance of bone scanning in ECD, because the scintigraphic pattern of involved skeletal sites may in themselves lead to the diagnosis. Several differential diagnoses are considered. The importance of bone scintigraphy as an imaging method in patients with an unclear diagnosis is discussed, as exemplary in ECD, as is its role for the detection of sites of skeletal involvement in other diseases.	10836686

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2000 Jun	5: Hum Pathol;31(6):734-9	Erdheim-Chester disease: evidence for a disease entity different from Langerhans cell histiocytosis? Three cases with detailed radiological and immunohistochemical analysis.	Kenn W, Eck M, Allolio B, Jakob F, Illg A, Marx A, Mueller-Hermelink HK, Hahn D	Department of Radiology, University of Wurzburg, Germany.	In this study, 3 cases of Erdheim-Chester disease were followed up over years and examined in detail both radiologically and immunohistochemically. All 3 cases showed the pathognomonic skeletal features for EC disease as well as an identical immunohistochemical phenotype quite different from LCH. Macrophages and Touton cells reacted strongly positive with the histiocytic marker CD 68, whereas staining with S100 and CD1a, markers for Langerhans cells, were negative. Both the immunohistochemical phenotype and the bone changes were clearly distinct from LCH.	10872668
2000 Jun	6: J Rheumatol;27(6):1550-3	"Coated aorta": a new sign of Erdheim-Chester disease.	Serratrice J, Granel B, De Roux C, Pellissier JF, Swiader L, Bartoli JM, Disdier P, Weiller PJ	Service de Médecine Interne, Hôpital de la Timone, Marseille, France.	We describe 3 unusual cases of Erdheim-Chester disease with periaortic fibrosis involving the whole aorta and leading to a "coated aorta" appearance on computed tomography scans. Faced with such a singular "coated aorta," bone scintigraphy can be very helpful when searching for Erdheim-Chester disease.	10852289
2000 May	7: AJR Am J Roentgenol;174(5):1327-31	Pulmonary involvement with Erdheim-Chester disease: radiographic and CT findings.	Wittenberg KH, Swensen SJ, Myers JL	Department of Diagnostic Radiology, Mayo Clinic, Rochester, MN 55905, USA.	We retrospectively reviewed the radiologic images of 15 patients with biopsy-proven Erdheim-Chester disease. Nine patients had chest radiographic images and CT scans that were available for review. Six men and three women were studied (age range, 25-70 years; mean age, 56 years). CONCLUSION: The most common findings of Erdheim-Chester disease with pulmonary involvement include an interstitial process characterized by smooth interlobular septal thickening and centrilobular nodular opacities, fissural thickening, and pleural effusions. On CT, six of nine patients had pericardial fluid and thickening or extrathoracic soft-tissue masses. Such findings are characteristic of Erdheim-Chester disease with pulmonary involvement. Definitive diagnosis requires correlating skeletal findings and lung biopsy findings.	10789787
2000 May	8: Nihon Kokyuki Gakkai Zasshi;38(5):380-4	[Erdheim-Chester disease presenting with pulmonary lesion]	Nakano H, Yano S, Kobayashi K, Kawasaki Y, Mikami M, Shishido S, Fukuda M, Kawabata Y	Department of Respiratory Medicine, Matsue Hospital, Shimane, Japan.	A 49-year-old man first visited our hospital in 1991 for further examination of abnormal pulmonary shadows. A chest radiograph and computed tomographic (CT) scan showed diffuse reticular shadows in both lung fields. The findings from a transbronchial lung biopsy specimen were not conclusive. Although there was little change in the abnormal pulmonary shadows, the patient's lung functions gradually deteriorated, indicating an obstructive defect. The patient was admitted in 1998 with the chief complaint of increasing dyspnea on exertion. A thoracoscopic lung biopsy specimen revealed proliferation of histiocytes with fibrosis in the pleura and perivascular interstitium. Immunohistochemically, the histiocytic cells were CD68-positive, alpha 1-antichymotripsin-positive, S100 protein-negative, and CD1a-negative. A bone scintigram and magnetic resonance images showed symmetrical diametaphyseal bone lesions in the distal femurs and the proximal tibiae; however, the epiphyses were spared. These findings were consistent with Erdheim-Chester disease. This is the first reported case of Erdheim-Chester disease with pulmonary involvement in Japan.	10921285

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2000 Apr	9: Skeletal Radiol;29(4):227-30	Erdheim-Chester disease with intramuscular lipogranuloma.	Yamamoto T, Mizuno K	Department of Orthopaedic Surgery, Kobe University School of Medicine, Japan.	We report on a rare manifestation of Erdheim-Chester disease with intramuscular lipogranuloma. The patient was a 66-year-old man who noted a soft tissue mass in the right quadriceps femoris muscle. Radiographs revealed symmetrical osteosclerosis in the diametaphysis of both femora and tibiae. An open biopsy revealed a proliferation of lipid-laden histiocytes in the femoral bone marrow and the quadriceps femoris muscle. To our knowledge, this is the second case of Erdheim-Chester disease involving muscle.	10855472
2000	10: J Comput Assist Tomogr;24(2):281-3	Pseudotumoral bilateral involvement of the breast in Erdheim-Chester disease: CT appearance.	Ferrozzi F, Bova D, Tognini G, Zuccoli G	Istituto di Scienze Radiologiche, Università degli Studi, Parma, Italy.	We report a case of pseudotumoral involvement of the breast in Erdheim-Chester disease. CT shows an enlargement of both breasts with inhomogeneous structure, microcalcifications, and foci of fatty density.	10752893
2000	11: Arch Orthop Trauma Surg;120(1-2):112-3	Erdheim Chester disease: a rare cause of knee and leg pain.	Sistermann R, Katthagen BD	Orthopädische Klinik, Städtische Kliniken Dortmund, Klinikzentrum Mitte, Germany. dr.si@t-online.de	A case of Erdheim Chester disease in a 51-year-old Turkish patient is described. Erdheim Chester disease is a rare form of lipoid granulomatosis. Knee and leg pain are the most common symptoms, and physicians working in orthopaedics and traumatology are the first to be consulted. Our patient demonstrated a typical bilateral, symmetric sclerosis of the metaphyseal region of long bones of the lower extremity, histologic examination revealed foamy, lipid-loaded histiocytes. The patient also suffered from arterial hypertension, diabetes insipidus and exophthalmos of the left eye. The diagnosis was confirmed by a bone biopsy, and the patient was treated with non-steroidal anti-inflammatory drugs, corticosteroids and vincristine.	10653118
2000 Oct	12: Nephron;86(2):195-6	Nephrotic syndrome and amyloid A amyloidosis in a patient with Erdheim-Chester disease.	Enríquez R, Cabezuelo JB, Martínez M, Sáez J, Sirvent AE, Amorós F, Reyes A			11014995
2000 Aug	13: Neuroradiology;42(8):625	Erdheim-Chester disease: a sinonasal lesion mimicking rhinoscleroma.	Marsot-Dupuch K, Le Hir P			10997572
2000 Aug	14: Nuklearmedizin;39(5):N72-3	Epiphyseal involvement in Erdheim-Chester disease: radiographic and scintigraphic findings in a case with lytic lesions.	Ruíz-Hernández G, Tajahuerce-Romera GM, Latorre-Ibáñez MD, Vila-Fayos V, Lara-Pomares A	Servicio de Medicina Nuclear, Hospital Provincial Castellon, Spain.		10984893
2000 Mar	15: AJR Am J Roentgenol;174(3):875-6	Erdheim-Chester disease involving bilateral lower extremities: MR features.	Kushihashi T, Munechika H, Sekimizu M, Fujimaki E	Showa University, School of Medicine, Tokyo, Japan.		10701649

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
2000	16: Eur Neurol;43(4):242-4	Erdheim-Chester disease with spinal cord manifestations.	Pego-Reigosa R, Brañas-Fernández F, Martínez-Vázquez F, Rivas-Bande MJ, Sanjuanbenito L, García-Villanueva M, Cortés-Laíño JA	Neurology Department, Hospital Xeral-Calde, Lugo, Spain. rpegor@medynet.com		10828658
2000	17: Clin Imaging;24(2):64-7	Perinephric xanthogranulomatosis: CT diagnosis and confirmation by CT-guided percutaneous biopsy.	Scheer M, Hon M, Fruauff AA, Blumenfeld W, Grossman ZD, Katz DS	Department of Radiology, Winthrop-University Hospital, 259 First Street, Mineola, NY 11501, USA.	Xanthogranulomatosis is an idiopathic, rare process in which lipid-laden histiocytes may deposit in various locations in the body, which if systemic is called Erdheim-Chester disease. A rare case of isolated retroperitoneal, bilateral perinephric xanthogranulomatosis is reported. The diagnosis was suspected on cross-sectional imaging and was confirmed by CT-guided percutaneous core biopsy.	11124472
2000 Jan	18: Joint Bone Spine;67(1):71-4	Intraosseous xanthoma without lipid disorders. Case-report and literature review.	Boisgard S, Bringer O, Aufauvre B, Joudet T, Kemeny JL, Michel JL, Levai JP	Department of Orthopedic Surgery, hôpital G. Montpied, CHU Clermont-Ferrand, France.	A case of intraosseous xanthoma in a patient with a normal lipid profile is reported. Hyperlipidemia is present in most patients with xanthomas. Intraosseous xanthomas are rare, particularly in normolipidemic patients, in whom the presenting symptom is pain without skin lesions. A lytic lesion with a rim of sclerosis is seen on radiographs. Histology shows foam cells, giant cells, and fibrosis. Intraosseous xanthoma is a benign tumor, and other diagnoses must be ruled out (histiocytosis X, Erdheim Chester disease, clear cell carcinoma metastasis). Surgical excision of the lesion is the elective treatment.	10773972
1999 Sep	1: Hum Pathol;30(9):1093-6	Chester-Erdheim disease: a neoplastic disorder.	Chetritt J, Paradis V, Dargere D, Adle-Biassette H, Maurage CA, Mussini JM, Vital A, Wechsler J, Bedossa P	Department of Pathology, Hospital de Bicêtre, France.	Chester-Erdheim disease is a rare non-langerhans cell histiocytosis characterized by a xanthomatous infiltration of foamy macrophages. The cause and pathogenesis remain unclear. The aim of the present study was to determine whether Chester-Erdheim disease is a polyclonal reactive disease or a clonal neoplastic disorder. The clonal status of samples obtained from five patients with Chester-Erdheim disease was studied. DNA was extracted from fixed and paraffin-embedded sections after microdissection and clonal status was studied using the Xchromosome inactivation pattern of the human androgen receptor gene (HUMARA assay). One patient was homozygous for the HUMARA gene and noninformative. Three other cases were monoclonal. One was polyclonal, and this case showed a dense reactive infiltrate in association with spumous macrophages. This study suggests strongly that Chester-Erdheim disease is a monoclonal lesion consistent with neoplastic disorder. Thus, Chester-Erdheim disease may be considered as the "macrophage" counterpart of Langerhan's cell histiocytosis in the histiocytosis spectrum. Further studies are needed to establish the origin of this clonal proliferation.	10492045

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
1999 Aug	2: Neth J Med;55(2):76-9	A patient with diabetes insipidus and periorbital swellings; Erdheim-Chester disease.	van der Lee I, Slee PH, Elbers JR	Department of Internal Medicine, St. Antonius Hospital, CM Nieuwegein, The Netherlands.	Erdheim-Chester disease is a rare multisystem disease in which a progressive xanthogranulomatous infiltration of several tissues can be seen. We describe a woman, known to have diabetes insipidus for ten years, with periorbital, retroperitoneal, mediastinal, axillar and inguinal involvement. On histological examination a granulomatous infiltration of fatty tissue and striated muscle was seen, consisting of Touton giant cells, histiocytes with foamy cytoplasm and lymphocytes. Immunohistochemical staining with CD-1a and S-100 was negative and on electron microscopy no Langerhans granules were seen. These findings led to the diagnosis of Erdheim-Chester disease. She had a good response on steroids. Because of some similar clinical features of Langerhans cell histiocytosis and Erdheim-Chester disease, a histiocyte disorder seems the most probable cause.	10474276
1999 Jun	3: J Korean Med Sci;14(3):323-6	Erdheim-Chester disease: a case report.	Park YK, Ryu KN, Huh B, Kim JD	Department of Pathology, College of Medicine, Kyung Hee University, Seoul, Korea. damia@chollian.net	A 42-year-old man with Erdheim-Chester disease (EC) is presented. This is the first case of this disease reported in Korea. The patient complained of knee pain and plain roentgenogram of the bilateral legs revealed diffusely increased density, coarsened trabecular pattern, and cortical thickening in the diaphysis, and metaphysis as well as epiphysis. Magnetic resonance imaging revealed that the lesions showed low signal intensity on T1-weighted images and heterogeneously low and high signal intensity on T2-weighted images. Histological examination of the biopsy specimen showed a xanthogranulomatous lesion consisting aggregations of foamy histiocytes and Touton-type giant cells. Immunohistochemical staining showed positive reaction to anti-S-100 and lysozyme in the cytoplasm of the giant cells.	10402177
1999 Jun	4: Orbit;18(2):99-104	Erdheim-Chester disease: a bilateral orbital mass as an indication of systemic disease.	Amrith S, Hong Low C, Cheah E, Oo Tan Y	Consultant Ophthalmologists, Mount Elizabeth Medical Center, Singapore	A case of bilateral orbital mass and xanthelasma of the eyelids is presented. Histology confirmed it to be a form of histiocytosis, possibly an Erdheim-Chester disease. This was further confirmed by the presence of a retroperitoneal mass and hydronephrosis, which resolved with treatment. A review of the literature on and pathological features of this rare fatal disease is presented.	12045992
1999 Apr	5: Presse Med;28(14):738-40	[Urinary complications of Erdheim-Chester disease]	Karsenty G, André M, Rossi D	Service d'Urologie, hôpital Salvator, Marseille.	BACKGROUND: Erdeheim-Chester disease is an uncommon histiocytosis. Fifty-nine cases have been reported in the literature. Bone lesions are usually inaugural followed by multiorgan involvement combining bone disease, orbital infiltration, diabetes insipidis and retroperitoneal infiltration. CASE REPORT: A 53-year-old man had Erdheim-Chester disease which progressed over 11 years. The patient developed extrinsic obstruction of the upper urinary tract. This unusual complication of Erdheim-Chester disease raised a difficult therapeutic problem as percutaneous drainage was impossible. The patient was treated with an endoprothesis allowing urine derivation. Surgical ureterolysis was avoided. DISCUSSION: Data in the literature favor use of minimally invasive endourological treatment for patients with urinary tract complications of Erdheim-Chester disease.	10230410
1999 Apr	6: Clin Nucl Med;24(4):252-5	Determination of extent and activity with radionuclide imaging in Erdheim-Chester disease.	Franzius C, Sciuk J, Bremer C, Kempkes M, Schober O	Department of Nuclear Medicine, University Hospital, Westfälische Wilhelms-Universität, Münster, Germany.	Erdheim-Chester disease usually involves the diaphyseal and metaphyseal regions of tubular bones and various visceral organs. A 56-year-old woman presented with the histologically confirmed diagnosis of Erdheim-Chester disease. A Tc-99m MDP bone scan revealed the entire extent of the skeletal disease and showed unusual involvement of the epiphyses and axial skeleton. In addition to MRI, a Ga-67 citrate scan including SPECT showed extensive soft-tissue infiltration of different organs. Both Tc-99m MDP and Ga-67 scintigraphy are useful tools in determining the distribution of this rare disease.	10466521

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
1999 Jan	7: Am J Surg Pathol;23(1):17-26	Erdheim-Chester disease: clinical, radiologic, and histopathologic findings in five patients with interstitial lung disease.	Egan AJ, Boardman LA, Tazelaar HD, Swensen SJ, Jett JR, Yousem SA, Myers JL	Department of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, Minnesota 55905, USA.	Erdheim-Chester disease is a clinicopathologic entity defined by a characteristic pattern of symmetric osteosclerosis caused by an infiltrate of mononuclear cells that include prominent numbers of foamy histiocytes. About half of patients have extraskeletal manifestations, including involvement of the hypothalamus/posterior pituitary, orbit, retroperitoneum, skin, lung, and heart. Pulmonary involvement is an uncommon but important manifestation of Erdheim-Chester disease because it causes significant morbidity and mortality. A review of the Mayo Clinic files produced four patients with confirmed Erdheim-Chester disease in whom lung biopsy had been performed. One additional patient was included from the University of Pittsburgh. Four patients were women. The mean age was 53.6 years (range 25-70 years). All patients had bilateral and symmetric sclerotic bone lesions characteristic of Erdheim-Chester disease, although in three the skeletal abnormalities were discovered only after lung biopsy. Four patients had dyspnea, and one also had a dry cough. One patient died 17 months after diagnosis. Chest radiographs showed diffuse interstitial infiltrates in all patients, with an upper zone predominance in three. Thoracic computed tomography (CT) scans showed thickening of the visceral pleura and interlobular septa with patchy associated fine reticular and centrilobular opacities and ground glass attenuation. Lung biopsy specimens showed an infiltrate of foamy histiocytes, lymphocytes, and scattered Touton giant cells with associated fibrosis in a striking lymphatic distribution. The infiltrate involved visceral pleura, interlobular septa, and bronchovascular bundles. Immunohistochemical stains were positive for CD68 in all cases and S-100 protein in four cases. Stains for CD1a were consistently negative. Ultrastructural studies in one case showed no Birbeck granules. Although in bone the histologic features of Erdheim-Chester disease may overlap with Langerhans' cell histiocytosis, its expression in the lung is distinct. Lung involvement in Erdheim-Chester disease has emerged as a unique radiographic and histologic entity.	9888700
1999	8: Clin Exp Pathol;47(2):71-6	Brain stem infiltration by mixed Langerhans cell histiocytosis and Chester-Erdheim disease: more than just an isolated case?	Vital C, Bioulac-Sage P, Tison F, Rivel J, Begueret H, Gomez C, Leaute-Labreze C, Diard F, Vital A	Laboratoire de Neuropathologie, Université Victor Segalen, Bordeaux, France.	Langerhans cell histiocytosis is classically considered as totally different from Chester-Erdheim's disease which consists in the infiltration of various parenchymas by macrophagic CD68-positive histiocytes. We report the case of a 46-year-old woman with a long history of diabetes insipidus who presented typical lesions of Langerhans cell histiocytosis on vulvar and skin biopsies as well as bony cellular infiltrates characteristic of Chester-Erdheim's disease. A few months later she presented cerebellar disorders and died after an 18-month course. At autopsy the pons was enlarged, due to numerous cellular infiltrates which were also scattered in the middle cerebellar pedoncles, dentate nuclei, midbrain and hypothalamus. There were S100-protein positive Langerhans cells intermingled with numerous ovoid CD68-positive histiocytes. There are a few reported cases of Chester-Erdheim's disease presenting foci of Langerhans cells histiocytosis in other parenchymas. In addition, there are 10 reported cases with diabetes insipidus and bilateral infiltration of the brain stem and cerebellum, considered as presenting either one type of histiocytosis or the other. Our case demonstrates that both histiocytoses may coexist in the brain and thus correspond in fact to the same pathology in certain particular cases.	10398577

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
1999	9: Eur Radiol;9(1):153-8	Erdheim-Chester disease: a case report and literature overview.	Kenn W, Stäbler A, Zachoval R, Zietz C, Raum W, Wittenberg G	Institut für Röntgendiagnostik der Universität Würzburg, Joseph Schneider Strasse 2, D-97080 Würzburg, Germany.	Erdheim-Chester (EC) disease belongs to the group of lipoid granulomatosis. Symmetric sclerosis of the meta- and diaphysis of long tubular bones are pathognomonic radiologic changes. Additionally, other skeletal segments can be affected. Extraskeletal manifestations can occur in almost all organs; lungs, pericardium, retroperitoneum, skin, and orbita play particularly important roles. The last case of 38 cases of Erdheim-Chester disease with an extraordinary mediastinal and perirenal involvement is described. For the second time following the initial description by Chester, an axial skeletal pattern of eburnated vertebra is shown.	9933400
1999 Jan	10: J Neurol Neurosurg Psychiatry;66(1):72-5	Neurological manifestations of Erdheim-Chester disease.	Wright RA, Hermann RC, Parisi JE	Department of Neurology, Mayo Clinic and Mayo Foundation, Rochester, Minnesota, USA. wright.russell@mayo.edu	Erdheim-Chester disease is a rare sporadic systemic histiocytic disease of unknown aetiology that affects multiple organ systems. The case records of all patients with Erdheim-Chester disease who had been seen at the Mayo Clinic between 1975 and 1996 were reviewed to assess the neurological manifestations of the disease. Two of 10 patients had neurological involvement. A 42 year old woman developed central diabetes insipidus and a progressive cerebellar syndrome. Brain MRI showed a lesion in the left pons with patchy gadolinium enhancement and T2 weighted signal abnormalities extending into both cerebellar peduncles and the medulla. Biopsy of the brainstem mass showed a xanthogranulomatous lesion. The second patient was a 53 year old man with retroperitoneal fibrosis secondary to xanthogranulomatous infiltration. Although he had no neurological symptoms and a normal neurological examination, MRI of the head showed multiple uniformly enhancing extra-axial masses along the dura of both convexities and the falx, and a mass in the left orbital apex. Both patients had the characteristic radiographic and bone scan findings of Erdheim-Chester disease. Review of the literature disclosed a wide variety of neurological manifestations in Erdheim-Chester disease. The most frequent CNS manifestations are diabetes insipidus, cerebellar syndromes, orbital lesions, and extra-axial masses involving the dura.	9886456
1999 Dec	11: J Urol;162(6):2084-5	Bilateral hydronephrosis in a patient with Erdheim-Chester disease.	Droupy S, Attias D, Eschwege P, Hammoudi Y, Benoit G, Jardin A	Department of Urology, Paris-Sud University School of Medicine and Bicêtre Hospital, Le Kremlin Bicêtre, France.		10569576
1999 May	12: Radiol Med (Torino);97(5):422-4	[Erdheim Chester disease: autonomous entity or expression of Langerhans cell histiocytosis? Report of a case]	Popolizio T, De Serio A, Scarabino T, Bisceglia M, Cammisa M	Dipartimento di Diagnostica per Immagini, IRCCS Casa Sollievo della Sofferenza, Rotondo FG. dallapiccola@operapadrepio.it		10432980
1999 Apr	13: Eur J Radiol;30(1):70-4	Erdheim-Chester disease.	Lozano JG, Lopez-Negrette L, Sanchez JL, Sala J	Servicio de Radiologia, Hospital Valle del Nalon, Asturias, Spain.		10389016

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1999 Jul	14: Hum Pathol;30(7):770-80	Posttraumatic fibro-osseous lesion of rib.	McDermott MB, Kyriakos M, Flanagan FL	Department of Radiology, Washington University School of Medicine, St Louis, MO, USA.	Eleven cases are described of an unusual, benign, fibro-osseous lesion of rib previously reported under a variety of designations, including painless fibro-osseous lesion resembling osteoid osteoma, symmetrical fibro-osseous dysplasia, focal Erdheim-Chester disease, and fibro-osseous pseudotumor. All patients were adults, most of whom were asymptomatic, the lesion discovered by bone scans done to rule out metastatic disease. A single rib was involved in eight patients and multiple ribs in three. A roentgenographic abnormality was apparent in only five patients. Histologically, all lesions showed a bland fibrous stroma in which resided an anastomosing network of bone trabeculae, having a zonal pattern of maturation from metaplastic woven to mature lamellar bone, with or without an associated xanthomatous component. Seven patients had a history of previous trauma, three with fractured ribs. Considering the relative infrequency of solitary rib lesions attributable to metastatic disease, it is proposed that in most cases there is no need for a diagnostic rib resection for these incidentally discovered, posttraumatic reparative lesions.	10414495
1998 Dec	1: J Intern Med;244(6):529-35	Endocrine manifestations of Erdheim-Chester disease (a distinct form of histiocytosis).	Tritos NA, Weinrib S, Kaye TB	Division of Endocrinology, Beth Israel Deaconess Medical Center, Boston, Massachusetts, USA.	Erdheim-Chester disease (ECD) is a disorder of unclear aetiology, characterized by exuberant histiocyte proliferation and a variable clinical course. We report the case of a woman with multi-organ involvement secondary to ECD. Central diabetes insipidus (CDI), hyperprolactinaemia, gonadotropin insufficiency and decreased insulin-like growth factor I levels were present, suggesting hypothalamic-pituitary dysfunction. The high-intensity signal of the posterior pituitary on T1-weighted images was absent on magnetic resonance imaging, but no sellar mass lesions or stalk thickening were apparent. Additionally, our patient had bilateral adrenal enlargement. Even though ECD is a rare cause of neuroendocrine dysfunction or adrenal enlargement, it should be considered in patients with these disorders in the setting of multiorgan disease.	9893107
1998 Oct	2: J Neurol Neurosurg Psychiatry;65(4):597-9	Cerebral Erdheim-Chester disease: report of two cases with progressive cerebellar syndrome with dentate abnormalities on magnetic resonance imaging.	Pautas E, Chérin P, Pelletier S, Vidailhet M, Herson S	Department of Internal Medicine, Hôpital de la Pitié-Salpétrière, Paris, France.	Two patients with Erdheim-Chester disease with progressive cerebellar dysfunction and pyramidal signs are reported on. Cerebral MRI showed bilateral increased signal intensity in peridentatal regions on T2 weighted sequences. Both patients had kidney and bone involvement, established on bone biopsy for one. One patient improved with steroid therapy. This contrasts with previous reports, which describe rare neurological manifestations and the failure of different therapeutic approaches.	9771797
1998 Jun	3: Orbit;17(2):97-105	Bilateral orbital involvement in Erdheim-Chester disease.	de Palma P, Ravalli L, Grisanti F, Rossi A, Marzola A, Nielsen I	Department of Ophthalmology, University of Ferrara, Ferrara, Italy	Erdheim-Chester disease is an idiopathic condition characterized by a xanthogranulomatous process infiltrating the bones, lungs, heart, retroperitoneum and other tissues. This condition is often fatal. Ocular findings are rare. The authors report a case of bilateral xanthelasmas and bilateral massive orbital infiltration in a 61-year-old man with severe retroperitoneal fibrosis, renal and cardiovascular problems. The ophthalmic manifestations and differential diagnosis of this peculiar pathologic condition are discussed.	12048709

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1998 May	4: Ann Dermatol Venereol;125(5):335-8	[Langerhans-cell histiocytosis and Erdheim-Chester disease: probably not a fortuitous association]	Boralevi F, Léauté-Labrèze C, Tison F, Bioulac-Sage P, Vital C, Delbrel X, Cony M, Géniaux M	Clinique Dermatologique, Hôpital Pellegrin, Bordeaux.	BACKGROUND: Erdheim Chester disease (MEC) is a rare non-Langerhans cell histiocytosis characterized by multi-visceral involvement. We report a case of MEC associated with Langerhans cell histiocytosis (HCL). CASE REPORT: A 46-year-old women presented skin and vulvar localization of HCL associated with typical MEC bone involvement. Despite chemotherapy (vinblastine) and prednisone, the disease progressed to involve the central nervous system, leading to fatal outcome. Post-mortem examination showed HCL in skin, MEC in bones and central nervous system, and intermediate histiocytic proliferation in the encephalon. DISCUSSION: Usually, MEC and HCL are considered as distinct entities. MEC is characterized by a xanthogranulomatous proliferation of CD 68+ foamy histiocytes nested in fibrosis, and HCL by a proliferation of PS 100+ and CD1a+ Langerhans cells. However, our observation, as well as previous reports, suggests that MEC is part of the HCL spectrum.	9747283
1998 May	5: Mov Disord;13(3):576-81	Erdheim-Chester disease with extensive intraaxial brain stem lesions presenting as a progressive cerebellar syndrome.	Evidente VG, Adler CH, Giannini C, Conley CR, Parisi JE, Fletcher GP	Department of Neurology, Mayo Clinic Scottsdale, Arizona 85259, USA.	We report a rare case of Erdheim-Chester disease (ECD) presenting as a progressive cerebellar syndrome and diabetes insipidus. On magnetic resonance imaging, a 7-mm extraaxial, enhancing mass was seen enveloping the right vertebral artery and was confirmed at autopsy to represent an adventitial xanthoma with lipid-laden, foamy histiocytes. The cerebellar syndrome most likely resulted from extensive histiocytic infiltration of the pons, particularly the basis pontis and middle cerebellar peduncles.	9613758
1998 Mar	6: Skeletal Radiol;27(3):127-32	Erdheim-Chester disease: radiographic findings in five patients.	Bancroft LW, Berquist TH	Department of Diagnostic Radiology, Mayo Clinic Jacksonville, FL 32224, USA.	We present the case histories of five patients with Erdheim-Chester disease, a rare lipoidosis that has several typical radiographic features. In all the patients, the diaphyses and metaphyses of the extremities demonstrated a symmetric pattern of diffuse or patchy increased density, a coarsened trabecular pattern, medullary sclerosis, and cortical thickening. The epiphyses were spared in four patients and partially involved in one. The axial skeleton was involved in one patient. Radiotracer 99mTc accumulated in areas of radiographic abnormalities in all patients. In one patient, MRI demonstrated an abnormal signal, corresponding to radiographic abnormalities. The signal was hypointense to muscle on T1-weighted sequences and heterogeneously hyperintense and hypointense to normal bone marrow on T2-weighted sequences. Xanthogranulomatous lesions infiltrated the retroperitoneum in one patient, the testes in one patient, the eyelids in one patient, and the orbits in two patients.	9554002
1998 Feb	7: Am J Respir Crit Care Med;157(2):650-3	Erdheim-Chester disease: a primary macrophage cell disorder.	Devouassoux G, Lantuejoul S, Chatelain P, Brambilla E, Brambilla C	Department of Respiratory Medicine, Hopital Albert Michallon, Grenoble, France.	Erdheim-Chester disease (ECD) is a rare focal or systemic infiltrative disorder resulting from xanthogranulomatous tissue deposition. Usually, bone marrow involvement affects long bone metaphyses symmetrically, but it spares the epiphyses. Retroperitoneal space, periaortic area, skin, and brain involvement have been described. Pulmonary involvement is frequent, occurring in 20% of cases. Reported histologic features in the lung include an infiltration of so-called lipid-laden macrophages and granulomatous lesions with fibrosis. Lung function outcome is unpredictable, but terminal respiratory failure is the most frequent cause of death. No effective treatment strategies have been described. We report a new case with lung and bone involvement occurring in a symptomatic woman. Histologic and electron microscopic analysis of the pulmonary infiltrate showed abnormal macrophages devoid of lipids forming nodular granulomas and rendering the previous hypothesis of this disease as a primary lipid storage disorder unlikely. These findings suggest that ECD histogenesis is instead based on a primary macrophage cell disease.	9476885

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1998 Apr	8: Rofo;168(4):397-9	[Erdheim-Chester lipogranulomatosis with involvement of the breast]	Jacobs A, Jäger H, Walther L, Schatz T	Institut für Strahlendiagnostik, Städtische Kliniken Dortmund Mitte.		9589106
1998 Mar	9: J Neurol Neurosurg Psychiatry;64(3):420-1	Erdheim-Chester disease and slowly progressive cerebellar dysfunction.	S Bohlega			9527179
1998 Nov	10: Dtsch Med Wochenschr;123(45):1337-42	[Xanthoma disseminatum with marked mucocutaneous involvement]	Tietge UJ, Maschek H, Schneider A, Gawehn AE, Wagner S, Manns MP, Schmidt HH	Abteilung Gastroenterologie und Hepatologie, Medizinische Hochschule Hannover.	HISTORY AND CLINICAL FINDINGS: When aged 23 years, a now 36-year-old man was first diagnosed as having xanthomas on the upper arms and shoulders. Xanthomas then progressed, affecting both the skin and the laryngo-pharyngeal mucosa. They were so marked that several laser-surgical interventions for their removal in the phayngo-laryngeal tract were necessary to ensure unimpaired breathing. There were also extensive confluent symmetrical cutaneous xanthomas over the upper and lower arms, the face, neck and trunk. Xanthomas and scars in the pharynx and larynx necessitated marked nasal breathing. INVESTIGATIONS: There was no laboratory evidence of abnormal lipid metabolism. The concentrations of cholesterol, triglycerides, lipoprotein (a), apolipoprotein A-1, apolipoprotein B, apolipoprotein E phenotype and steroles were all normal. The biochemical composition of LDL, VLDL and HDL particle was also unremarkable. Histological examination of resected xanthomas revealed dense infiltrations of the interstitial spaces by foam-cell histiocytes with multiple lipid vacuoles, typical of xanthoma disseminatum. TREATMENT AND COURSE: Neither probucol nor cholesterol synthesis enzyme inhibitors nor glucocorticoid medication influenced the xanthomas. The only effective treatment was removal of the most unsightly or obstructing lesions. But the sars left removal in the mucocutaneous regions caused obstruction in the laryngopharyngeal tract. CONCLUSION: The cause of xanthoma disseminatum remains unknown. Skeletal muscle can also be extensively infiltrated. This case shows similarities to Erdheim-Chester disease, another are xanthomatous condition.	9835892
1997 Dec	1: Neurology;49(6):1702-5	Cerebral manifestation of Erdheim-Chester disease: clinical and radiologic findings.	Bohlega S, Alwatban J, Tulbah A, Bakheet SM, Powe J	Department of Neurosciences, King Faisal Specialist Hospital & Research Centre, Riyadh, Saudi Arabia.	A 33-year-old woman presented with a 3-year history of progressive numbness in the hand, cerebellar ataxia, limb weakness, nystagmus, and dysarthria. T2-weighted MRI revealed abnormal foci of increased signal intensity mimicking demyelinating plaques in the periventricular white matter, and brain 18FDG-PET scan showed increased uptake in the pons. Biopsy from a tibial lesion showed aggregates of foamy histiocytes in the intertrabecular spaces replacing the bone marrow, characteristic of Erdheim-Chester disease. The patient was treated with craniospinal radiation. After 6 months, the clinical picture was stable and the MRI was unchanged.	9409372

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1997 Nov	2: Arch Ophthalmol;115 (11):1467-8	A case of Erdheim-Chester disease with orbital involvement.	Valmaggia C, Neuweiler J, Fretz C, Gottlob I	Department of Strabismus and Neuro-ophthalmology, Kantonsspital, St Gallen, Switzerland.	The Erdheim-Chester disease is a rare idiopathic, systemic, histiocytic disorder. To our knowledge, ocular involvement has been reported in only 16 cases. We describe a 55-year-old man who had symmetrical exophthalmos and several skin nodules on the arms and trunk. A magnetic resonance imaging scan confirmed the presence of bilateral, intraconal, retrobulbar tumors. An examination of the histopathologic features of orbital and skin biopsy specimens revealed xanthogranulomatous infiltrate with Touton giant cells. Further systemic investigations showed bone and retroperitoneal involvement. Three years later, multiple eyelid xanthelasmas developed in the patient. These findings are consistent with the diagnosis of the Erdheim-Chester disease. The patient's condition is stable under therapy with low-dose corticosteroids. His survival is longer than usually described in the literature.	9366683
1997 Nov	3: J Neuropathol Exp Neurol;56(11):1207-16	Pathology of the central nervous system in Chester-Erdheim disease: report of three cases.	Adle-Biassette H, Chetritt J, Bergemer-Fouquet AM, Wechsler J, Mussini JM, Gray F	Département de Pathologie (Neuropathologie) Hôpital Universitaire Henri Mondor, Créteil, France.	Chester-Erdheim disease is a rare form of non-Langerhans cell histiocytosis consisting of disseminated xanthogranulomatous infiltration and fibrosis that primarily involves the bones, visceral organs and systemic fatty spaces. Involvement of the central nervous system is variable, and neuropathological features have seldom been documented. We report the neuropathological findings in 3 autopsy cases. One patient had radiological and pathological bone changes characteristic of Chester-Erdheim disease. Neuropathology revealed multiple characteristic xanthogranulomas disseminated in the cerebral hemispheres, hypothalamus, cerebellum, and brainstem. The second patient presented first with cutaneous lesions characteristic of Langerhans cell histiocytosis. She subsequently developed bone abnormalities suggestive of Chester-Erdheim disease, which was confirmed by autopsy, raising the possibility of a common spectrum of histiocytosis including both diseases. Gross examination of the brain was normal, however, microscopy showed infiltration of the brain by characteristic non-Langerhans cell xanthogranulomas. The third patient presented with systemic features characteristic of Chester-Erdheim disease. Neurological signs included gait disturbance, seizures and confusion. Examination of the brain did not show any histiocytic infiltration, but did show changes suggestive of Hallervorden-Spatz syndrome. Association of Chester-Erdheim disease and Hallervorden-Spatz syndrome has not been previously reported. The relationship between both conditions is unclear.	9370231
1997 Oct	4: Metabolism;46(10):1215-9	Erdheim-Chester disease: low low-density lipoprotein levels due to rapid catabolism.	Schmidt HH, Gregg RE, Shamburek R, Brewer BH, Zech LA	Molecular Disease Branch, National Heart, Lung, and Blood Institute, National Institutes of Health, Bethesda, MD, USA.	We have identified a 44-year-old patient with symmetrically excessive xanthomatosis, called Erdheim-Chester disease (ECD), and simultaneously decreased levels of low-density lipoprotein (LDL) cholesterol. Clinically, this patient presents lipoidgranulomatosis of numerous long and flat bones with involvement of the liver, spleen, pericardium, pleura, thyroid, skin, conjunctiva, and gingiva. However, the patient does not have any signs of atherosclerosis. So far, the underlying defect has not been elucidated. We performed a LDL-apolipoprotein B (apoB) kinetic study in the ECD patient and a normal control to determine the etiology of the low LDL level in ECD. LDL was isolated from both subjects, radioiodinated with either 131I or 125I, and injected simultaneously into the ECD patient and the normal control. Normal and ECD LDL was catabolized at the same rate after injection into the control subject (fractional catabolic rate [FCR], 0.43/d and 0.46/d, respectively). Therefore, LDL isolated from an ECD subject is metabolically normal. In contrast, autologous LDL injected into the ECD subject showed a markedly increased catabolism (FCR, 0.69/d) compared with that in the control subject (FCR, 0.43/d). This is the first report about increased catabolism of LDL cholesterol in a patient.	9322810

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1997	5: Clin Imaging;21(5):328-31	Erdheim-Chester disease demonstrated by bone radiograph and scans.	Kim EE, Romero JA	Department of Nuclear Medicine, University of Texas M. D. Anderson Cancer Center, Houston 77030, USA.	Two cases of patients with Erdheim-Chester disease (EC) are presented with interesting scintigraphic findings. Differential diagnosis of bone scan and radiographic findings is briefly discussed.	9316751
1997 May	6: J Neurosurg;86(5):888-92	Erdheim-Chester disease of the central nervous system. Report of two cases.	Babu RP, Lansen TA, Chadburn A, Kasoff SS	Department of Neurosurgery, New York Medical College, Valhalla, USA.	The authors report two cases of Erdheim-Chester disease (ECD), an illness of unknown pathogenesis. Generally, this disease process involves the metaphyseal and diaphyseal portions of the long bones, the lungs, and the retroperitoneum; however, other tissues may be involved including the central nervous system (CNS). To date only two cases of CNS-related ECD have been reported. The present report adds to the literature by documenting two more recent cases of ECD involving the CNS. The clinical presentations of these cases, their radiological findings with special reference to magnetic resonance imaging, pathological determination, and clinical management are briefly reviewed.	9126908
1997 Apr	7: Histopathology;30(4):353-8	Erdheim-Chester disease with prominent pulmonary involvement associated with eosinophilic granuloma of mandibular bone.	Kambouchner M, Colby TV, Domenge C, Battesti JP, Soler P, Tazi A	Service d'Anatomie Pathologique, Hopital Avicenne, Bobigny, France.	We report a patient with eosinophilic granuloma localized to the left mandible who was subsequently shown to have Erdheim-Chester disease involving the lower extremities, omentum and lung. The diagnosis of eosinophilic granuloma was based on the presence of typical CD1a+ Langerhans' cell granulomas in a biopsy of mandible. The diagnosis of Erdheim-Chester disease was established on the basis of the pattern of radioisotopic uptake by long bones, seen on a technetium bone scan, and the presence of characteristic histopathological features in biopsies of lung and peritoneum. The pathological findings in lung were compatible with the abnormalities observed by tomodensitometry, but strikingly different from those seen in Langerhans' cell granulomatosis. The differences in the histological features of pulmonary involvement seen in the two diseases, and the possible relationship between Langerhans' cell granulomatosis and Erdheim-Chester disease, are discussed.	9147084
1997	8: J Fr Ophtalmol;20(5):331-2	[Erdheim-Chester disease. Survey of a rare non-Langerhans histiocytosis]	Veyssier-Belot C, Wechsler J, Cacoub P			9238468
1997 Nov	9: Klin Monatsbl Augenheilkd;211(5):342-4	[Bilateral adult periocular xanthogranuloma]	Spraul CW, Grossniklaus HE, Lang GK	L. F. Montgomery Eye Pathology Laboratory, Emory University School of Medicine Atlanta, GA, USA.	PATIENT: A 62-year-old woman was evaluated for a bilateral subconjunctival mass that had been present for 6 months. With magnetic resonance imaging the lesion could not be delineated form the lacrimal glands. A biopsy was performed and histologic examination exhibited a xanthogranulomatous lesion with multiple giant-cells of the Touton type. The differential diagnosis of the adult xanthogranuloma is Erdheim-Chester disease, necrobiotic granuloma, xanthoma, Langerhans histiocytosis, and Rosai-Dorfman syndrome. CONCLUSION: Hyperlipemia should be excluded, in addition, in the presence of necrobiosis, paraproteinemia.	9527593

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1997 Jun	10: Am J Surg Pathol;21(6):664-8	Breast involvement by extranodal Rosai-Dorfman disease: report of seven cases.	Green I, Dorfman RF, Rosai J	Department of Pathology, Memorial Sloan-Kettering Cancer Center, New York, New York 10021, USA.	Seven cases of breast involvement by extranodal Rosai-Dorfman disease are presented. The patients were women and their ages ranged from 15 to 84 years. Three patients had disease confined to the breast; one had involvement of the breast and ipsilateral axillary lymph nodes, and two had bilateral breast involvement as well as disseminated systemic disease. In all cases the clinical and radiographic presentation of the breast lesion raised the possibility of a malignant tumor. All but one of the lesions were treated by excisional biopsy. Microscopically, the lesions were relatively circumscribed, often multinodular masses, located in the breast stroma, with or without associated involvement of the subcutaneous tissue and dermis. They were composed of sheets of S-100 protein-positive large histiocytes displaying lymphocytophagocytosis, scattered in a polymorphous background of mature lymphocytes and plasma cells. The microscopic differential diagnosis includes idiopathic granulomatous mastitis, infective granulomas, Langerhans' cell histiocytosis, Erdheim-Chester disease, fibrous histiocytoma, and malignant melanoma.	9199644
1996 Dec	1: J Radiol;77(12):1213-21	[Imaging of Erdheim-Chester disease]	Gomez C, Diard F, Chateil JF, Moinard M, Dousset V, Rivel J	Service de Radiologie A, Professeur Diard, Hôpital Pellegrin, Bordeaux.	Erdheim-Chester disease is a form of Histiocytosis which involves the adults and is distinct from Histiocytosis X. It is characterized by a constant diaphyseal and metaphyseal bone involvement predominating in the lower links. The diagnosis can readily be envisaged when the typical radiological findings are present. Bone involvement may be isolated and well tolerated, or can be associated with systemic involvement and a severe prognosis. We describe three cases of women aged 46, 50 and 73 years. One patient presented with isolated bone lesions, while the two others had a multiorgan localization. From the three cases and from an extensive review of the literature, we describe the spectrum of bone and visceral lesions that can be seen by imaging. The emphasis is put on lesions of the skeletal system, the retroperitoneum, the nervous system, and the pericardium. Furthermore, the relationships between Erdheim-Chester disease and Histiocytosis X are discussed.	9033881
1996 Aug	2: Nippon Igaku Hoshasen Gakkai Zasshi;56(9):681-3	[A case report of Erdheim-Chester disease]	Furutani K, Kurosawa Y, Kageyama T, Kaneko M	Department of Radiology, Seirei Hamamatsu General Hospital.	Erdheim-Chester disease is a rare and distinctive lipid granulomatosis with characteristic pattern of radiographic changes in bone. The characteristic radiographic finding is an unusual symmetrical sclerosis at the diametaphyseal portions of many long bones. This study demonstrates a case of Erdheim-Chester disease and mainly documents radiographic findings.	8831229

Publ Date	Publication	Title	Author(s)	Author Contact	Editted Abstract	PMID
1996 May	3: Medicine (Baltimore);75(3):157-69	Erdheim-Chester disease. Clinical and radiologic characteristics of 59 cases.	Veyssier-Belot C, Cacoub P, Caparros-Lefebvre D, Wechsler J, Brun B, Remy M, Wallaert B, Petit H, Grimaldi A, Wechsler B, Godeau P	Service de médecine interne, hôpital Pitié-Salpêtrière, Paris, France.	We made a retrospective evaluation of clinical and radiologic features, treatment, and outcome of Erdheim-Chester disease, a rare non-Langerhans cell histiocytosis. We had 7 patients coming from 3 French teaching hospitals and reviewed 52 cases from the literature. These cases were considered to have Erdheim-Chester disease when they had either typical bone radiographs (symmetrical long bones osteosclerosis) and/or histologic criteria disclosing histiocytic infiltration without features for Langerhans cell histiocytosis (no S-100 protein, no intracytoplasmic Birbeck granules). Ages at diagnosis ranged from 7 to 84 years (mean +/- SD = 53 +/- 14 yr) with a male/female ratio of 33/26. Bone pain was the most frequent clinical sign (28/59), mostly located in the lower limbs. Exophthalmos and diabetes insipidus were found in respectively 16/59 and 17/59 patients. General symptoms (fever, weight loss) and "xanthomas" (mainly located on the eyelids) were present in 11/59 patients. Retroperitoneal involvement was found in 17/59 patients. Skeletal X-ray showed typical osteosclerosis of the diaphysis of the long bones in 45/59 patients. Bone radiographs showed osteolytic lesions of the flat bones (skull, ribs) in 8 patients. Histologic diagnosis was performed after a bone biopsy (28 patients), a retroorbital biopsy (9 patients), and/or a biopsy of the retroperitoneal infiltration or the kidney (11 patients). Six of our 7 patients but only 5 of 52 patients from the literature had the complete histologic criteria, disclosing no Birbeck granules or S-100 immunostaining. In other cases, histologic results usually described a xanthogranulomatous infiltration by foamy histiocytes nested in fibrosis. Treatment was corticotherapy (20/59), chemotherapy (8/59), radiotherapy (6/59), surgery (3/59) and immunotherapy (1 patient). Twenty-two patients died after a mean follow-up of 32 +/- 30 mo (range, 3-120 mo). In conclusion, Erdheim-Chester disease may be confused with Langerhans cell histiocytosis as it sometimes shares the same clinical (exophthalmos, diabetes insipidus) or radiologic (osteolytic lesions) findings. However, it also appears to have distinctive features. Patients are older and have a worse prognosis than those with Langerhans cell histiocytosis, and the diagnosis relies on the association of specific radiologic and histologic findings.	8965684
1996 Jan	4: Hum Pathol;27(1):91-5	Erdheim-Chester disease: a case report with immunohistochemical and biochemical examination.	Ono K, Oshiro M, Uemura K, Ota H, Matsushita Y, Ijima S, Iwase T, Uchida M, Katsuyama T	Department of Pathology, Tosei General Hospital, Seto, Japan.	This report describes a 47-year-old man with Erdheim-Chester disease (EC), the second case reported in Japan. The patient complained of knee pain, and the roentgenogram of the bilateral legs revealed symmetric osteolytic lesions with sclerosis of the metaphyseal regions of the long bones. Histological examination of the biopsy specimen showed a xanthogranulomatous lesion consisting of aggregations of foamy macrophages and Touton-type giant cells. Immunohistochemical study of the foamy cells in the lesion showed positive reaction to anti-Kp-1, anti-S-100 alpha, beta, anti-neuron-specific enolase (NSE), anti-alpha-1-antichymotrypsin, anti-alpha-1-antitrypsin, and anti-lysozyme antibodies. Electron microscopy showed many lipid droplets in the cytoplasm, but no Langerhans granules. These results suggested that the disease was part of the spectrum of histiocytosis but was different from Langerhans cell histiocytosis. Biochemical analysis of material extracted from a lesion showed the predominance of cholesterol ester. The disease progressed to central diabetes insipidus, and the involvement of multiple organs was indicated by a magnetic resonance image.	8543320

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1995 Oct	1: AJNR Am J Neuroradiol;16(9):1787-90	Erdheim-Chester disease: MR of intraaxial and extraaxial brain stem lesions.	R Martinez	Department of Radiology, New York Medical College, Valhalla, USA.	A case of Erdheim-Chester disease demonstrates cerebral hemispheric involvement, as well as and intraaxial and extraaxial brain stem involvement in a patient with symptoms of paraparesis, urinary incontinence, visual loss, ataxia, vertigo, proptosis, and nystagmus. Persistent gadopentetate dimeglumine enhancement was noted in the extraaxial cervicomedullary brain stem lesion 23 days after injection. However, the supratentorial lesions fail to show similar persistent enhancement. This case also demonstrates MR features characteristic of retrobulbar infiltration.	8693976
1995 Apr	2: AJNR Am J Neuroradiol;16(4):735-40	Neuroradiologic aspects of Chester-Erdheim disease.	Caparros-Lefebvre D, Pruvo JP, Rémy M, Wallaert B, Petit H	Department of Neurology, CHRU Lille, France.	In three cases of histologically proved Chester-Erdheim disease there was a large anterior epidural lesion from C-3 to L-2 in one patient; dural masses and orbital infiltration in a second patient; and dural, choroid plexus, retroorbital, and hypophyseal lesions in a third patient. Diabetes insipidus, exophthalmia, long bone lesions, and retroperitoneal infiltration were present.	7611030
1995 Feb	3: J Neurol Neurosurg Psychiatry;58(2):238-40	Erdheim-Chester disease and slowly progressive cerebellar dysfunction.	Fukazawa T, Tsukishima E, Sasaki H, Hamada K, Hamada T, Tashiro K	Hokuyukai Neurology Hospital, Sapporo, Japan.	A 59 year old woman developed pronounced thirst, increased water intake, and increased urinary output followed by slowly progressive cerebellar symptoms. Brain MRI showed abnormal hyperintensity on T2 weighted studies in the region of both dentate nuclei without atrophy of the cerebellum or the brainstem. A 99mTC diphosphonate bone scan showed bone lesions in the distal parts of both femurs as well as distal and proximal parts of both tibias. The diagnosis of Erdheim-Chester disease was made by bone biopsy. This is the first case of Erdheim-Chester disease presenting as a slowly progressive cerebellar syndrome and diabetes insipidus, and also showing high signal lesions in deep cerebellar nuclei on MRI. Skeletal surveys are indicated for patients with otherwise unexplained slowly progressive cerebellar symptoms.	7876861
1995	4: Ann Pathol;15(1):59-62	[Erdheim-Chester disease. Clinico-pathologic study of two cases]	Farre I, Copin MC, Boulanger E, Remy J, Wallaert B, Gosselin B	Service d'Anatomie et de Cytologie Pathologiques, Hôpital Calmette, CHU Lille.	Erdheim-Chester disease is a rare visceral xanthogranulomatosis characterized by bilateral, symmetrical sclerosis of the metaphyseal regions of long bones and infiltration of foamy, lipid-laden histiocytes. Clinically, it ranges from an asymptomatic, focal process to a fatal, systemic disease. We report two new cases, different in their presentation and extension.	7702670
1995	5: J Fr Ophtalmol;18(3):220-5	[Orbital Erdheim-Chester disease]	Offret H, Hannouche D, Frau E, Doyon D, Quillard J, Schaison G	Service d'Ophtalmologie, C.H.U. Bicêtre.	Erdheim-Chester disease is related to a tissue infiltration of foamy histiocytes. Results of immunoperoxydase stains for S-100 and T6 protein, the Langerhans cells antigen, is negative. It is a multisystemic disease, and it particularly involves bones and orbit. The visual prognosis is threatened, and the disease may lead to a fatal issue. Treatments have poor effects on the disease. Patients sometimes have good symptomatic response to corticotherapy. This case was revealed by headaches and diabetes insipidus. The orbital infiltration was asymptomatic.	7759761
1995 May	6: AJR Am J Roentgenol;164(5):1115-7	Erdheim-Chester disease involving breast and muscle: imaging findings.	Tan AP, Tan LK, Choo IH	Department of Diagnostic Radiology, National University of Singapore.	7717216	
1994 Aug	1: Am J Surg Pathol;18(8):843-8	Retroperitoneal xanthogranuloma in a patient with Erdheim-Chester disease.	Eble JN, Rosenberg AE, Young RH	Indiana University School of Medicine, Indianapolis.	A case of Erdheim-Chester disease with retroperitoneal and renal sinus xanthogranuloma that occurred in a 50-year-old woman is presented. The 12 previously reported cases of Erdheim-Chester disease associated with retroperitoneal xanthogranuloma are reviewed and compared with 13 sporadic cases of retroperitoneal xanthogranuloma. Retroperitoneal xanthogranuloma is distinguished from inflammatory malignant fibrous histiocytoma by its lack of neutrophils, inconspicuous vascularity, lack of nuclear atypia, and abundant collagen. It is distinguished from inflammatory fibrosarcoma by its numerous foamy histiocytes, relative lack of plasma cells, and lack of nuclear atypia; it is distinguished from retroperitoneal fibrosis principally by its many foamy histiocytes, lack of plasma cells, and lack of vasculitis.	8037299

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1994	2: J Fr Ophtalmol;17(3):200-3	[Erdheim-Chester disease. A rare etiology of retrobulbar tumor]	Chollet P, Eyremandi R, Lesueur L, Arne JL	Service d'Ophtalmologie, Hôpital Purpan, Toulouse.	The authors report the case of a 47 year old man who presented with bilateral retro-ocular tumor and an inflammatory syndrome as the first sign of his disease. Later on, the illness became polyvisceral and biopsies of retro-ocular and retro-peritoneal tissues revealed the diagnosis of Erdheim-Chester disease. The patient died a few months later.	8182258
1994	3: J Comput Assist Tomogr;18(3):503-5	T of Erdheim-Chester disease presenting as retroperitoneal xanthogranulomatosis.	Chiang KS, Larson TS, Swee RG, Bostwick DG, LeRoy AH	Department of Radiology, Mayo Clinic, Rochester, MN.		8188927
1993 Oct	1: Rev Rhum Ed Fr;60(9):601-9	[Erdheim-Chester disease: report of a case, review of the literature and discussion of the relation to Langerhans-cell histiocytosis]	Pertuiset E, Laredo JD, Lioté F, Wassef M, Jagueux M, Kuntz D	Clinique de Rhumatologie, Hôpital Lariboisière, Paris.	Erdheim-Chester disease is an endogenous, non-genetically-determined lipidosis characterized by infiltrates of foamy, lipid-laden histiocytes and by bilateral symmetric foci of sclerosis in appendicular long bones. The clinical spectrum ranges from focal bone lesions to systemic disease with life-threatening visceral involvement. In one third of patients, roentgenograms show focal osteolysis within areas of sclerosis. Authors report a new case of Erdheim-Chester disease documented by two bone biopsies in different sites. Features in their patient included: 1) osteolysis and sclerosis of the long bones of the limbs and maxillas, with CT scan evidence of cortical rupture; 2) on magnetic resonance imaging studies, heterogeneous foci of decreased signal intensity on T1 images and heterogeneous areas of moderately increased signal intensity on T2-weighted images; 3) increased serum osteocalcin levels; 4) laboratory evidence of chronic inflammation with no extraosseous manifestations. The clinical, radiological, and pathological features of Erdheim-Chester disease are different from those of Langerhans cell histiocytosis. However, three cases of patients with both conditions have been reported in the literature, suggesting that there may be links between the two diseases.	8012336
1993 May	2: J Clin Pathol;46(5):481-2	Erdheim-Chester disease with epiphyseal and systemic disease.	Athanasou NA, Barbatis C	Department of Pathology, Nuffield Orthopaedic Centre, Oxford.	A case of Erdheim-Chester disease which affected the epiphysis and showed evidence of systemic disease is presented. Clinical and histopathological similarities with other forms of disseminated Langerhans' cell histiocytosis are noted, particularly reaction of infiltrating histiocytes for S100 and HLA-DR.	8320335
1993	3: Trans Am Ophthalmol Soc;91:99-125; discussion 125-9	Periocular xanthogranulomas associated with severe adult-onset asthma.	Jakobiec FA, Mills MD, Hidayat AA, Dallow RL, Townsend DJ, Brinker EA, Charles NC	Massachusetts Eye and Ear Infirmary, Boston.	This article describes six patients who presented, usually bilaterally, with yellow-orange, elevated, indurated, and nonulcerated xanthomatous eyelid lesions, typically extending into the anterior orbital fat, and sometimes involving the extraocular muscles and the lacrimal gland. Because the eyelids remained intact and because the process did not reach the deep orbital and perioptic connective tissues, visual acuity was well preserved. There is cosmetic morbidity and occasionally motility restriction with advancing involvement of the extraocular muscles. All patients had variably severe adult-onset asthma that required treatment with systemic prednisone and inhalants. No evidence of Erdheim-Chester disease was found in any patient, but the appearance in one patient, after 25 years of follow-up, of a separate subcutaneous necrobiotic xanthogranulomatous lesion in the mandibular region with an associated paraproteinemia, suggests that at least some of our cases might be a mild form of necrobiotic xanthogranuloma. For this reason, we would suggest repeated periodic serum protein immunoelectrophoretic studies as well as evaluation for lymphoma. Therapy probably should consist of low doses of periorbital radiotherapy coupled with high doses of corticosteroids. Should this not be successful, then systemic administration of corticosteroids with chemotherapeutic agents might be efficacious, as in necrobiotic xanthogranuloma.	8140711

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1992 Oct	1: Presse Med;21(36):1714-6	[Chester-Erdheim's disease. A case]	Boulanger E, Talaszka A, Le Monies de Sagazan H	Service de Néphrologie-Hémodialyse, Hôpital Victor Provo, Roubaix.	We report the 32nd case of a multivisceral form of Erdheim-Chester disease. This exceptional pathology is a diffuse xanthogranulomatosis which comes within the scope of histiocytosis. The originality of this case is due to cerebral localizations and to the fact that some symptoms have been observed for a long time: diabetes insipidus, exophthalmos and stubborn intertrigo.	1480576
1992 Oct	2: Radiol Med (Torino);84(4):471-5	["Erdheim-Chester" disease. Description of a case]	Serafini F, Carcello A, Viglietta G, Poggi C, Mandreoli M	Servizio di Radiologia, Ospedale Policlinico S. Orsola-Malpighi, USL 28, Bologna.		1455035
1992 Sep	3: J Prosthet Dent;68(3):399-401	Implant rehabilitation in Erdheim-Chester disease: a clinical report.	Brahim JS, Guckes AD, Rudy SF	Clinical Investigations and Patient Care Branch, National Institutes of Health, National Institute of Dental Research, Bethesda, Md.	Successful osseointegration of endosseous titanium implants is thought to be dependent upon close apposition of bone to the implant surface. The integration of implants in this patient was achieved despite the lipid-laden histiocytic infiltration of the bone marrow. Presumably, enough unaffected stromal cells were present to allow sufficient bone formation for osseointegration of the implant fixtures. This result invites speculation regarding both the mechanism of osseointegration and the minimum surface area of bone-implant interface necessary for achieving and maintaining osseointegration of titanium implants. This patient is periodically examined to determine if the loaded fixtures will remain clinically immobile for a prolonged period.	1432751
1992	4: Skeletal Radiol;21(1):64-7	Case report 710: Symmetrical eosinophilic granuloma of the lower extremities (proven) and Erdheim-Chester disease (probable).	Strouse PJ, Ellis BI, Shifrin LZ, Shah AR	Department of Diagnostic Radiology, Henry Ford Hospital, Detroit, Michigan.	We present a case of symmetrical EG of the lower extremities in a 36-year-old man. Several entities are considered in the differential diagnosis. However, many of the features bear a striking resemblance to ECD, which probably coexists in this case. A link between the two entities, EG and ECD, has been suggested by others. Future experience may confirm this hypothesis.	1546341
1991 Jun	1: Arch Ophthalmol;109(6):850-4	Orbital and eyelid involvement with Erdheim-Chester disease. A report of two cases.	Shields JA, Karcioglu ZA, Shields CL, Eagle RC, Wong S	Ocular Oncology Service, Wills Eye Hospital, Philadelphia, PA 19107.	Erdheim-Chester disease is an idiopathic condition characterized by infiltration of the heart, lungs, retroperitoneum, bones, and other tissues by a fibrosing xanthogranulomatous process composed of xanthomatous histiocytes and Touton giant cells. This condition is often fatal, with death due to cardiomyopathy, severe lung disease, or chronic renal failure. Ocular findings with this potentially fatal disease are rare. We report the clinical and histopathologic findings in two cases of bilateral xanthelasmas and bilateral orbital infiltrates in association with Erdheim-Chester disease. The first patient was a 38-year-old man with cardiovascular and renal disease and severe retroperitoneal fibrosis. The massive orbital infiltration produced bilateral blindness. The second patient was a 77-year-old man with severe cardiovascular disease and retroperitoneal fibrosis. The diagnosis was confirmed in both patients with retroperitoneal and orbital biopsies. Both patients had the unusual occurrence of bilateral xanthelasmas with bilateral, diffuse orbital masses, eye findings that should alert the clinician to the possibility of this serious systemic disease.	2043074

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1991 Jun	2: Arch Pathol Lab Med;115(6):619-23	Erdheim-Chester disease. Case report with autopsy findings.	Fink MG, Levinson DJ, Brown NL, Sreekanth S, Sobel GW	Department of Pathology, Humana Hospital-Michael Reese, Chicago, Ill. 60616.	Erdheim-Chester disease is a rare pathologic entity characterized by symmetrical radiodensities in the metaphyseal and the diaphyseal portions of the long bones. Fibrosis, osteoblastic cortical bone deposition, and fibroxanthomatous granulomas with lipid-laden macrophages and multinucleated giant cells, which have a particular tropism for connective and adipose tissues, are the pathologic hallmarks. To our knowledge, 27 cases have been reported in the literature since the entity was first described in 1930. Protean clinical features range from a focal and asymptomatic process to a multisystemic infiltrative disease. We describe the clinical course of a new case and review the extensive pathologic findings at autopsy, including those demonstrated by light and electron microscopy and cytochemical and immunocytochemical studies.	2039348
1991 Jun	3: Radiologe;31(6):307-9	[Cerebral manifestations of Erdheim-Chester disease]	Kujat C, Junk B, Hermes M, Martin J, Dewes W	Funktionsbereich Kernspintomographie, Universität des Saarlandes, Homburg/Saar.	Cerebral manifestations of Erdheim-Chester disease are variable, giving a picture like that of multiple sclerosis. White matter lesions are located mainly in cerebellum and pons and lipid granulomas in the meninges. An asymptomatic lesion in the choroid plexus, with prolonged uptake of Gd-DTPA is described for the first time.	1882073
1991 Jun	4: Radiologe;31(6):297-306	[Erdheim-Chester disease]	Kujat C, Martin J, Püschel W	Neuroradiologisches Institut, Universität des Saarlandes, Homburg/Saar.	Erdheim-Chester disease (ECD) is characterized by lipid granuloma in the long tubular bones, which leads to pathognomonic symmetrical sclerosis of their metaphyses and diaphyses. Lipid granuloma may also be present in numerous other mesenchymal tissues, especially lung, orbit and retroperitoneal space. The clinical course and prognosis of the disease depend on these lesions. Reviewing 30 cases published since 1931 and a personal case with S100 positive cells, we present the typical radiological and clinical findings. There is striking resemblance to chronic disseminated histiocytosis X.	1882072
1991 Apr	5: Presse Med;20(13):607	[A rare cause of exophthalmos, Erdheim-Chester disease]	Sellami M, Sellami F			1827907
1991	6: Am J Pediatr Hematol Oncol;13(1):42-6	A xanthogranulomatous histiocytosis in a child presenting with short stature.	Globerman H, Burstein S, Girardina PJ, Winchester P, Frankel S	Department of Pediatrics, New York Hospital-Cornell Medical Center, New York.	We evaluated a 7-year-old boy presenting with a neck mass that was diagnosed as juvenile xanthogranuloma on excisional biopsy. Despite this diagnosis, an exhaustive evaluation was undertaken because of marked short stature. Examination revealed growth hormone deficiency and diabetes insipidus, as well as widespread lesions in the head, mediastinum, retroperitoneum, skeleton, and elsewhere. Biopsies of the lesions in the mediastinum and right tibia suggested a diagnosis of xanthoma disseminatum with bony involvement, suggesting the Erdheim-Chester variant of xanthogranulomatous histiocytosis, previously reported only in adults. The diagnosis is contrasted to the more common clinical entities of juvenile xanthogranuloma and the Langerhans' cell histiocytoses. This case illustrates the gravity with which otherwise unexplained short stature should be considered.	1903027
1990 Sep	1: Oral Surg Oral Med Oral Pathol;70(3):294-6	Premature alveolar bone loss in Erdheim-Chester disease.	Valdez IH, Katz RW, Travis WD	National Institute of Dental Research, National Institutes of Health, Bethesda, Md.	Erdheim-Chester disease is a rare histiocytosis also known as lipoid granulomatosis. Oral findings have not been reported previously to our knowledge. This case report documents evidence of oral sequelae of Erdheim-Chester disease. A patient whose course was followed for 10 years at the National Institutes of Health had premature alveolar bone resorption. He underwent full-mouth extraction at age 29 years because of severe periodontitis. Histopathologic evidence of Erdheim-Chester disease was demonstrated in the periodontal soft tissues. In the ensuring years, accelerated resorption of the residual ridges precluded the use of conventional dentures. We recommend early preventive dental management for patients with Erdheim-Chester disease.	2216355

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1990	2: Eur J Nucl Med;16(1):55-60	Scintigraphic findings and follow up in Erdheim-Chester disease.	Sandrock D, Merino MJ, Scheffknecht BH, Neumann RD	Department of Nuclear Medicine, Warren G. Magnuson Clinical Center, National Institutes of Health, Bethesda, MD 20892.	Two cases of Erdheim-Chester disease are presented: a 26-year-old white male patient with lipoidgranulomatosis of numerous long and flat bones and infiltration of pericardium, pleura, liver, spleen, thyroid, skin, conjunctiva, gingiva, and false vocal cord; and a 54-year-old white male with involvement of bones, orbits, brain, pericardium, and retroperitoneum. The scintigraphic findings in this disease are described, and a comprehensive review of the 27 previously reported cases is given including an assessment of the value of scintigraphy for diagnosis and follow up of this rare disease.	2407535
1990	3: AJNR Am J Neuroradiol;11(6):1267-70	MR of diabetes insipidus in a patient with Erdheim-Chester disease: case report.	Tien R, Kucharczyk J, Newton TH, Citron JT, Duffy TJ	Neuroradiology Section, University of California, San Francisco 94143.		2124077
1990	4: Trans Pa Acad Ophthalmol Otolaryngol;42:931-7	Clinical spectrum of histiocytic tumors of the orbit.	Shields JA, Shields CL	Ocular Oncology Service, Wills Eye Hospital, Thomas Jefferson University, Philadelphia, Pennsylvania 19107.	Histiocytic tumors of the orbit comprise an unusual group of lesions characterized by the infiltration of the orbital tissues by xanthomatous cells. Recently, there have been a number of new observations regarding the various histiocytic tumors that can affect the orbit. The condition previously referred to as histiocytosis X is believed to represent a proliferation of Langerhans cells and the term Langerhans cell histiocytosis is often used instead of histiocytosis X. Juvenile xanthogranuloma has been demonstrated to affect the orbit without involving the skin or the iris. The Erdheim-Chester disease is a condition of adults characterized by infiltration of bone, retroperitoneum, heart, lungs and other tissues by xanthoma cells. This condition has recently been recognized to produce a classic ophthalmological picture of bilateral xanthelasmas and bilateral proptosis. The authors review their personal experience with several patients with histiocytic tumors of the orbit and stress the clinical spectrum of these conditions.	2084989
1989 Sep	1: Radiology;172(3):791-2	Cerebral Erdheim-Chester disease: persistent enhancement with Gd-DTPA on MR images.	Tien RD, Brasch RC, Jackson DE, Dillon WP	Department of Radiology, University of California San Francisco 94143.	A case of Erdheim-Chester disease with intracerebral masses containing characteristic lipid-laden histiocytes is presented. These unusual lesions remained enhanced on magnetic resonance images obtained 8 days after injection of gadolinium diethylenetriaminepentaacetic acid (DTPA) dimeglumine. Chemical analysis of a biopsy specimen revealed a high concentration of gadolinium. Findings suggest that the Gd-DTPA complex or possibly a gadolinium-containing metabolite may be retained for extended periods in this unusual type of histiocytic lesion.	2772189
1989 Mar	2: J Bone Joint Surg Am;71(3):456-64	Erdheim-Chester disease. A report of three cases.	Lantz B, Lange TA, Heiner J, Herring GF	Department of Orthopaedic Surgery, University of Arkansas for Medical Sciences, Little Rock 72205.		2925725
1989	3: Ann Dermatol Venereol;116(11):837-40	[Erdheim-Chester disease: a form of xanthogranulomatosis]	Wechsler J, Michaud V, Bagot M, Guillaume JC, Fraitag S, Brugières P, Brun B, Revuz J	Service d'Anatomie pathologique, Hôpital Henri-Mondor, Créteil.		2619181

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1988 Oct	1: Clin Nucl Med;13(10):736-41	Lipid granulomatosis: Erdheim-Chester disease.	Molnar CP, Gottschalk R, Gallagher B	Department of Radiological Sciences and Diagnostic Imaging, Foothills Hospital, Calgary, Alberta, Canada.	Twenty-six cases of lipid (cholesterol) granulomatosis, Erdheim-Chester Disease (ECD), have been described in the literature to date. A new case of ECD in a 33-year-old man with an unusual presentation of exudative ascites following a four year history of abdominal pain is reported. The radiographic and bone scan findings in this disease have been established and Ga-67 scan findings are reported. The Tc-99m sulphur colloid bone marrow and In-111 chloride scan findings are presented.	3180598
1988	2: Czech Med;11(1):57-64	A xanthogranulomatous process encircling large blood vessels (Erdheim-Chester disease?).	Mergancová J, Kubes L, Elleder M	School of Medicine, Charles University, Hradec Králové.	The case of a strange type of generalized xanthogranulomatosis, ending by a lethal kidney complication, is described in a woman aged 68. The clinical symptoms of the patient were not characteristic and did not lead to the actual diagnosis. The process was located along the basal brain arteries, it adhered to the adventitia of the descending thoracic aorta and of the coronary arteries. An identical xanthogranulomatous infiltrate was found in the peripelvic adipose tissue of both kidneys, where it led to stenosis of the proximal ends of both ureters; to a lesser extent such infiltrates appeared also in the periportal areas of the liver and in the bone marrow. Histological findings grant the possibility of the Erdheim-Chester disease. Differential diagnosis is subjected to discussion.	3133188
1988 Sep	3: Arthritis Rheum;31(9):1215-6	Erdheim-Chester disease associated with hydrocalycosis and arthropathy.	Brown R, van den Berg R, Hurst NP, Allen PW			3422024
1988 Apr	4: AJR Am J Roentgenol;150(4):869-71	Langerhans cell histiocytosis with the radiographic findings of Erdheim-Chester disease.	Waite RJ, Doherty PW, Liepman M, Woda B	Department of Radiology, University of Massachusetts Medical Center, Worcester 01605.		3258103
1988 Oct	5: Am J Clin Pathol;90(4):377-84	Xanthoma of bone.	Bertoni F, Unni KK, McLeod RA, Sim FH	Department of Diagnostic Radiology, Mayo Clinic, Rochester, Minnesota 55905.	The authors report on 21 cases of "primary" xanthoma of bone. Twenty of the patients were older than 20 years old. The male-female ratio was 2:1. The presenting symptom was pain in 13 patients and neurologic symptoms in 2; in 6 patients, the lesion was an incidental finding. All but one of the lesions in this series were solitary, and the flat bones (pelvis, rib, skull) were the most frequently involved sites. Radiographically, a well-defined, sometimes expansile lytic lesion, with either a small area of surrounding reactive bone or a distinct sclerotic margin, was seen. Microscopically, foam cells, giant cells, cholesterol clefts, and fibrosis were present in varying degrees. In none of these cases was there an identifiable underlying lesion. The differential diagnosis includes Erdheim-Chester disease (a multisystemic granulomatosis) and bone involvement in sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfman disease). More important is the differential diagnosis with metastatic clear cell carcinoma. Xanthoma of bone is a benign lesion, and complete or even partial removal is effective. Xanthomas may represent a "burnt-out" benign condition such as fibrous dysplasia or histiocytosis X.	3140652

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2006	Haematologica	**High-dose chemotherapy followed by autologous hematopoietic stem cell transplantation for adult histiocytic disorders with central nervous system involvement**	Nathalie Gaspar Pascaline Boudou Julien Haroche Bertrand Wechsler Eric Van Den Neste Khe Hoang-Xuan Zahir Amoura Remy Guillevin Julien Savatovski Nabih Azar Jean-Charles Piette Véronique Leblond	*From the Service d'hématologie clinique, Hôpital Pitié-Salpétrière, Paris, France (NG, PB,, NA, VL); Service de Médecine Interne, Hôpital Pitié-Salpétrière hospital, Paris, France (JH, BW, ZA, J-CP); Service d'hématologie, Cliniques Universitaires de Saint Luc, Brussel, Belgium (EVDN); Service de Neurologie, Hôpital Pitié-Salpétrière hospital, Paris, France (KH-X); Service de Neuroradiologie, Hôpital Pitié-Salpétrière hospital, Paris, France (RG, JS).*	We postulated that high-dose chemotherapy (HDC) followed by peripheral autologous hematopoietic stem cell transplantation might help to control refractory central nervous system (CNS) histiocytic disorders. Six patients with histiocytic CNS involvement were treated in this way. Two patients achieved non-active disease status, although one relapsed at 84 months. Two patients had regressive disease, one of whom progressed at 21 months. One patient had progressive disease at 14 months. One patient had extra-CNS progression but CNS regression. After a median follow-up of 22.4 months, only one of the six patients still has non-active disease. Treatment was effective on craniofacial and space-occupying brainstem lesions, and was ineffective on neurodegenerative lesions.	n/a