

# ECD Case Log

Case #	S e x	YOB	Dx date	Age at Sx onset	Age at ECD dx	Symptoms	ECD Involvement
1	M	1972	November-10	21	36	Lightheadness, Kidney issues in 1990's dx'd as multi cystic dysplastic kidney disease, controlled by meds. Chronic sinus problems. MRI showed masses behind the eyes. Surgery revealed a benign scar tissue type mass.	Lesions on the brain, mass behind eye, kidney, long bones of leg.
2	M	1940	7/7/2010 at death	45	66	1980's first symptoms were high blood pressure dx'd as renal artery stenosis, profuse perspiration, and itching/burning sensation after a shower. In 1999 had pain in knees, shins, lower back and flank dx'd as age related. In 2001 had night sweats, muscle cramps, anemia dx'd as myelofibrosis. In 2004 had extreme weight loss, extreme fatigue, nausea, numb upper lip, back rash, small, longstanding skin lesion bx'd, congestive heart failure, brain lesions, kidney failure, dialysis, fevers of unknown origin, kidney transplant. After transplant (2005 – 2007) had balance issues, slurred speech, loss of tooth material, extreme and urgent need to urinate, extreme pain in various parts of body, shortness of breath, double vision, excessive mucous in throat, swallowing issues, many infections. Surgery to remove mass on adrenal gland was aborted due to benign scar-like tissue around the vena cava. Adrenal gland biopsied 2 separate times. In late 2005 required feeding tube due to inability to swallow and loss of appetite. At the same time a superpubic catheter was placed due to inability to completely void bladder, dx'd as neurogenic bladder. In 2006 required stent in ureter of transplanted kidney due to stricture thought to be result of chronic UTI infections. Complete workups at Methodist in Houston, M.D. Anderson and Duke with no diagnosis.	Kidney, brain, adrenal gland, heart, lungs, bone, peritoneal area, testes, teeth, virtually entire body.
3	M	1970	June-05	25	36	Bone pain in right femur, diabetes insipidus. Diagnosed with LCH in 1995 after lesion removed from right femur. Inability to have children due to drugs. Lesion removed from skull in 2000. Kidney involvement in 2006. New biopsy of left pelvic bone led to ECD diagnosis.	Head (pituitary gland, temporal lobe), eyes, kidney, bones, adrenal gland, high blood pressure, tonic-clonic seizures
4	M	1956	April-10	46	50	Earliest symptoms, night sweats, fever, WBC High, RBC Low, overall cholesterol levels low, LDH high; C-reactive protein level from lowest of 57 to high exceeding 100. Thigh bone, knee, ankle pain, tired easily, w/ general feeling of malaise. Just prior to dx 4 yrs from onset of symptoms, had lost 25 pounds, was in Congestive Heart Failure, Fluid both lungs, weak, very anemic, in constant pain, fever, legs, ankles swelling. Had, MRI showed excessive lymph nodes, which led to abdominal surgery to take biopsies f/u with CT and CT Pet. Platelet issues required transfusions prior to surgery, steroids. Continues w/ Leg pain, entire skeleton, back pain and High blood pressure. .Osteoporosis, lower spine. Recently developed itchy spots. Update: 2010 Continues w/5MUI 3x/week Interferon; no new sites of involvement in last PET Scan (5-10); Pain on daily basis has been greatly reduced; continues with bad day following shot; pain comes on quickly over entire skeleton following stressful incident or day; hot weather has worsening effect on condition; itchy legs (red spots) increases, dehydrates quickly, loses energy or has none to begin with and spends most days "napping" after 10 hours of sleep, needs additional 4 hours during day. Caregiver has noticed memory loss as it relates to things happened in past 10 years or shorter time-frame. Since stabilizing disease, he has been able to reduce some medications, i.e., blood pressure. Now on low-dose Topral as a precautionary measure. Only sees doctor every three to six months as he is considered at "maintenance" levels. They are all too busy to look further into ECD.	Kidney, abdominal aorta, bone in lower back, neck and legs, mandible.

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Case #	Sex	YOB	Treatments Tried	Current Treatment	Current Status	Last Update Date
1	M	1972	See Current	Tamoxifen. Doctors are suggesting cytoxan because of past success with this drug. Patient hesitant to switch due to desire to have children.	Tired from tomoxifen. Night sweats sometimes.	4/10/2008
2	M	1940	Kidney transplant in 2004, anti-rejection drugs (prograf, cellcept, prednisone) from 2004 until 2007; inpatient methylpredisonole 1g/day x 5 day steroid treatment in 2005, IVIG treatment in 2007	n/a	Deceased 7/2007	7/12/2008
3	M	1970	Surgery to remove lesion in right femur (1995). Titanium rod placed in femur at that time to support the bone. (DDAVP for Diabetes Insipidus)? Surgery to remove tumor on skull (2000). Steroid injections and radiation to remove tumors behind the eyes (2001, 2002). Radiation for pituitary tumor (unsuccessful). Interferon (stopped in 11/06 after 8.5 mos. as he did not respond well to it and had severe side effects if he took other medication), chemotherapy (Vinblastine & prednisone - 3 treatments) helped shrink a bone tumor, but side effects were bad. Tried Methotrexate and Remicaid in 2008 but quit treatment due to side effects. Low dose Interferon again for 1 year. Now just Anakinra.	Anakinra, anti-seizure medications	Balance & speech problems (more pronounced during lower barometric pressure). Short term memory loss. Fatigue. Anxiety. Tonic-clonic seizures this summer.  Anakinra has taken away the bone pain. Able to stop narcotics and only use ibuprofen every so often.	9/18/2010
4	M	1956	Current only.	Interferon 5 MUI 3x week since diagnosis, plan is to remain on this long term; prednisone 5mg; Toprol XL & Altace for Blood Pressure; Prozac to negate side effects of Interferon (can be rage, suicidal) Update: 2010 Prednisone between 5-7mgs /day. Topral low dose; Paxil, antidepressant (anti-anxiety)	Doing well, disease slowed down on interferon. CT PET shows no more affected sites. Previous sites show no activity. Very tired from interferon, especially the day following the shot. Zero energy, takes lots of naps. Update:2010 Low or no energy; exhaustion; fevers; pain intermittant (following stress)	8/29/2010

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Case #	Sex	YOB	Dx date	Age at Sx onset	Age at ECD dx	Symptoms	ECD Involvement
5	M	1962	October-10	40	44	Poor balance and some slurring of speech and itchy spots. Had one major epileptic fit and some partial seizures so not allowed to drive. Epileptic seizure provoked more investigations. Previously had double vision, slurred speech and wobbliness. Brain MRI & removed longstanding skin lesion indicated EDC. Some night sweats. Lost weight while hospitalized in 2007. Odd sensations in the front part of the soles of feet (like numbness after having a jab although they aren't numb when they are touched). Problems with bladder needing to pass urine often and in a hurry (not believed to be diabetes insipidus). A PET scan after the last cladribine treatment had finished in Jan. 2009, showed a reduction in the activity of the disease. The PET will be repeated in 3 months. Going to Paris in early March to see Dr Haroche for assessment and for any advice he can offer, and to be part of his research project. Whilst attending Dr Tony Chu in London Cladribine was suggested. Got in touch with Professor Pete Hillmen in Leeds as it was much closer. Had subcut Cladribine for 5 consecutive days each month for 6 months. Well tolerated but Pet scan did not show improvement. Bad chest problems Winter 2009 (consolidated lungs > steroid treatment; respiratory syncytial virus pneumonia). Steroids continued for 6 months. Started low dose methotrexate (40mg once a week). PET scan shows improvement of most areas.	Brain, chest, skin, eyes and retroperitoneal area, Lungs
6	M	1964	June-10	43	43	2 months of sudden and intense illness. Initially thought food poisoning or stomach flu. Remained nauseated, began losing weight, gray colored stools. Three weeks later developed 104 degree fever. Extremely jaundiced. Then thought he had hepatitis. Entered into hospital via ER for 1 month hospital stay with liver, kidney, lung and heart failure. Lung biopsy resulted in major bleeding issues due to low platelets. Night sweats, but they were while on steroids.	Liver, kidney, lung, heart.
7	M	1961	July-10	40	46	2001-Diabetes insipidus, excessive urine output and unusual, constant fatigue. DDAVP, one prescrip, solved Diabetes insipidus. Brain surgery recommended, but refused. 2004-Onset of severe leg pain involving shins. Shrugged off pain as fatigue from constant walking on concrete & asphalt surfaces at work. 3/2007-Unusual fatigue returns enough to cause concern. 3-4/2007-Sought Family Physician's advice about leg pain and fatigue. Suspicious due to lack of follow up concerning Pituitary Xanthum, He referred me to a Neurosurgeon. MRI reveals the growth has increased. Neurosurgeon referred me to an Endocrinologist to explore effects of the growth on hormone levels. Testosterone is found to be dangerously low, and a hormone replacement gel is prescribed. A Transnasal biopsy is attempted, but fails due to depth of growth in brain. Neurosurgeon referred me to a Neural-Ophthalmologist, who successfully gains biopsy after Optical Ultrasound locates growth grazing Optic Nerve. Local lab in NC was unable to identify biopsy. Tissue sent to Washington University in St. Louis, MO. Growth ID'd as Xanthogranuloma. Referred by Neural Ophthalmologist to Oncologist, who stated Xanthums rarely grow in one area, and a Full Body Bone Scan is scheduled Xanthums discovered in numerous areas from the brain to the Tibias. Upon images being examined by Oncologist, a diagnosis of Erdheim-Chester Disease was made.	One growth, the first detected, begins surrounding Pituitary gland, continues down around Carotids and reaches back up into the orbits.  A shadow is seen in (right?) kidney.  Xanthum detected around the aorta in the Peritoneal (intestinal) cavity.  Area of the lower Femurs and entirety of Tibias are found to be involved.
8	M	1963	May-10	34	41	1997 noticed initial dizziness when climbing up high. Developed loud snoring in 2003 but not sleep apnea. Testing at that time showed high blood pressure. 2004: worsening balance & dizziness, slurred speech and emotional incontinence. CAT scan showed growth in kidney and puffiness around kidneys. Bone scan indicated ECD. Biopsy of kidney growth confirmed ECD. Currently chronic constipation, sexual problems. Diabetes insipidus since childhood due to injury to head. Stamina and strength have decreased, but mostly that is from the lack of physical activity. Balance and speech issues continue, but have adapted to them. (12/08 – speech and cognitive issues seem to be getting worse, but no pain issues)	Cerebellum-pons-pituitary area of brain, kidney 'halo' and lesion, femurs
9	M	1967?	October-10			Swollen eyes, achy joints, slurred speech, wobbly walk.	Lungs, eyes, nodes under jaw.

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Case #	Sex	YOB	Treatments Tried	Current Treatment	Current Status	Last Update Date
5	M	1962	Has been on (1) Interferon, then (2) azathioprine and then (3) cyclophosphamide and then back to (4) only interferon 3mu every other day and then (5) subcutaneous cladribine subcutaneously for 5 days consecutively at intervals of one month, for a total of 6 months beginning in July 2008 due to activity seen in bones for ECD. Prednisolone (reducing over 6 months) Methotrexate 40mg once a week	Lamotrigine 100mg bd (for epilepsy) Warfarin 7mg od (for pulmonary embolus) Co-valsartan od (for blood pressure) Fluoxetine 20mg od (for depression) Methotrexate 40mg once weekly	Retired at 46. Generally improved. Less slurry; only use sticks when I go out, stair lift only occasionally (after too much slurrp); only one pair of glasses needed for reading and distance; swallowing usually OK so the weight is piling on!; still coughing quite a lot. Restarted regular trumpet lessons, and doing voluntary sessions in local neurology ward and a local "resource centre for those with chronic neurological conditions". Also making bread, cakes, and jam at home.	8/17/2010
6	M	1964	Steroids, interferon.	Interferon 6 MIU 3x week. Prednisone 4 mg (tapering off). Plan to taper interferon to 3 MIU when off prednisone. Bone marrow stem cell transplant on hold because ECD is stable and risk of transplant.	Feels worn out from interferon, feels like he had the flu, but not too bad. Stable. On blood pressure medicine. Anemic. Has numbness in right leg, strange sensations on foot.	4/10/2008
7	M	1961	Androgel testosterone replacement gel, 5 g tube, did not absorb as quickly as Testim, per Endocrinologist. Vicodin (e) - Did nothing for leg pain. Caused drowsiness.	Intron A injections- 3 million units (.5 ml), 3 times per week. Testim testosterone replacement gel- 50 mg per 5 g tube, 4 times per week. Fentanyl, 12 mcg patch- replace patch every 72 hrs. *25 mcg patch replaced w/ 12 mcg due to pain relief lasting beyond 84 hrs. Bupropion 150 mg tablets- one per day (AM) for depression. Clonazepam 1 mg tablets- one per day (PM) for leg spasms. Loratadine 10 mg- One as needed to relieve itching in shins which began after Radiation treatments used to shrink Xanthums in that area. If symptoms return, I have qualified for financial assistance from Johnson & Johnson to acquire Leustatin, which has shown promise in treating the Xanthums. Ester-C- 1000 mg- One per day to boost bodies immune system. Vit. D w/ D3- One per day for energy during Winter and to support prostate health. Saw Palmetto 320 mg ( per 2 capsules)- Two per day for prostate health. Concern due to Testosterone replacement gel's side effect of Prostate enlargement. Fish Oil 1000 mg- 2 per day for brain health. Folic Acid 400 mcg- Helps w/ energy level and cell production, such as in the stomach, to aid w/ digestion. Calcium, Magnesium & Zin (Cal.-600+mg, Magn.- 250+ mg, Zinc- 10 mg)- Lab work showed Calcium levels to be low. Zinc is involved w/ eye health. Magnesium 250 mg- 2 per day to ease constipation caused by Fentanyl. Men's Multivitamin. Silicea terra- To relieve unusual body odor, from combination of meds and supplements, I assume.	Currently I feel great. My pain level is nearly non-existent. I do, at times, have trouble w/ balance and "steering", ie I often run into walls turning corners and veer right a bit when walking. I'm very far sighted and, more often than I'd like, fall asleep quickly during the day, sometimes for 2-3 hrs. This is very unusual for me. I walk for 30-60 minutes 5-7 days per week, work out w/ 20 lb dumbbells 3-5 days per wk, do a number of abdominal exercises, push-ups, flexibility routines, use an ellipse machine for 10 min.'s after walking 4-6 days/wk. It takes me 2-3 days. But, I push mow our roughly one acre, mountain yard and am working on an aesthetic plan for our yard. I may need rest regularly to get through the day. But, I will NOT give in and let someone else handle my responsibilities, and this disease doesn't have what it takes to damage my faith. NOBODY/NOTHING wins in a fight against God! He's in my corner. Thus, we win. ECD loses!	10/11/2010
8	M	1963	2CDA in 2005; stable since then.	Avapro & Norvasc for high blood pressure. Nortriptylene for emotional incontinence. DDAVP for diabetes incipidus. CPAP for sleep problems.	Stable since 2005. Some good days, some bad. Mayo clinic in April, 2008, showed no changes in scan for last 2 years.	7/9/2008
9	M	1967?	Remicaid and Methotrexate along with Cellcept	None since 11/06.	In remission since 3/08.	4/16/2008 (incomplete history)

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10	F	1967	June-05	37	39	Night sweats and leg pain. Tired and cold with intermittent fevers. Internal pressure inside head and optic nerves, treated with LPI to release pressure immediately and meds of Umerux and Prednisone .After 6 months of treatment pressure disappeared. Diabetes insipidus 3 years after onset of symptoms. From 2006 - 2008 ears plugged with yellowish discharge that must be removed once a month.	Kidney, bone, brain.
11	M	1952	June-05	44	46	Increased thirst (Nov 1995) 4 plus gallons of water per day. Low blood pressure, swelling (legs, face), Low grade fever (for 3-1/2 years) (1996). Shin pain (1997). Bone pain (1998). Slight reduction of tissue (abdominal, kidneys) after off of morphine. Last 7 years I've remained stable (no increased tissue). Learning to deal with fatigue through better nutrition. Control pain with brain wave stimulation.	Skeletal, Central diabetes insipidus, osteoporosis
12	M	1940	June-10	54	64	Began with pain in the long bones of the legs in about 1989. In 1/04 stomach began getting bigger and would feel full after eating only a few bites. In 12/03 began having chills and fever every evening. In 12/01 prior to knee surgery an MRI revealed what was thought to be prostate cancer. (Prostate had been removed in 1990. Prostate cancer was retreated in 1998 with radiation therapy and PSA was normal one month prior MRI of knee so return of prostate cancer was not considered a correct diagnosis.) Knee surgery in 01/2002 resulted in infection of the knee. In 04/02 began having problems with legs again and orthopedic surgeon wanted to do a biopsy of Rt. leg above and below knee. This resulted in having a bone biopsy of hip which resulted in an infection. In 03/04 fatigue and shortness of breath resulted in a consultation with an infectious disease doctor who ordered a CT of the pelvis, abdomen and chest. Within two days 2200 cc of fluid was removed from the sac surrounding the heart. 8 biopsies of stomach tumors were done with no definitive diagnosis except none malignant. A month later finally got a diagnosis.06/04	Masses in stomach, chest, and Behind eyes, involvement in bones skull, shoulders, arms ,Legs, & hips
13	F	1955	24 <sup>th</sup> Dec., 2005	41	48	1995: (possible sx's): large fibroids in uterus necessitating hysterectomy 1995, no dx of fibrosis at the time remembered: 1999: (probably ECD related?): severe 'heartburn' pains: dx'd as narrowing of the coronary artery supplying blood to the right atrium - a stent was implanted 1999 – an unusual cholesterol/ triglycerides ratio was noticed (very low HDL giving HDL/Triglycerides ratio that indicates very high likelihood of coronary artery disease – the material blocking the artery was said to be cholesterol but no biopsy was done. 2001: Definite ECD symptom: fluttering / waviness in peripheral vision – biopsy taken: dx: Acute Idiopathic Orbital Inflammatory Syndrome – fibrotic tumour growth around both optic nerves. The biopsy material was retained and used in later analysis. Treatment with cortisone (75mg/d for three months reducing to 10mg/d over three months before coming off) slowed growth but did not reduce size of tumours. Radiation treatment on both eyes had no effect. Treatment with Imuran reduced the size of tumours somewhat relieving pressure on the eyes – by 2004 the growths stabilised and vision returned to a reasonable level however glasses are now needed permanently and cortisone led to the onset of Type 2 diabetes. 2005: 'galloping in the ears' / very high blood pressure: scans showed a growth in the retroperitoneal area, adjacent to but not enclosing the aorta and invisible but probably enclosing and compressing both renal arteries (hence the high bp). Biopsy of retroperitoneal mass confirmed fibrotic tissue. A stent was inserted into the right renal artery, eliminating the blood pressure problem and treatment with Tamoxifen & Imuran has stabilised the tumours (no growth, no shrinkage) has been seen in over two years. 2006: Ophthalmologist did research work and initiated a review of all the available material including carrying out scans of the long bones: a firm dx of ECD was made, confirming the treatment regime as appropriate. Re-evaluation included both biopsies which were now interpreted as characteristic of ECD 2007: scan done which showed no change in the size of the tumours in 4 years even though the specialist in looking inside the eye had expected significant improvement. Lifelong (since 16) hypothyroidism. Lifelong difficulty with metabolising fat. Enamel on front teeth "breaks off."	Retro-ocular area surrounding both optic nerves; retroperitoneal area with two visible growths around renal arteries, probably fibrotic tissue related to that biopsied from the retroperitoneal area. Density changes/ lesions to all long bones (arms and legs).
14	M	1964	June-05	28	36	Initial problem was a small growth on pituitary gland discovered around 1992. Later when dizziness became an issue the growth on the pituitary gland was removed and as a result blindness occurred in right eye. Began taking testosterone injections and enjoyed a period of feeling good. In 1996 while in Michigan the symptoms returned and had another surgery to remove tissue from the brain stem and optic nerve. This resulted in balance issues, reduced sight in the left eye. The doctor did not understand what he was seeing and put the case on the computer. A doctor from Korea actually came up with the diagnosis. Previously had issues with colon disease, but now temporary colostomy has been removed.	Eyes, lung, kidney, brain
15	F	1950	May-00	38-40	49	At least ten years of severe hip/leg/knee pain. Can't remember exact date of onset. Was diagnosed during this time with fibromyalgia, chronic fatigue syndrome, lymphoma, arthritis. Diabetes Insipidus onset in 1990. Hyperthyroidism onset in 1998 but may/may not be connected. Significant weight gain during last fifteen years. Routine abdominal CT in 2000 showed retroperitoneal mass with extensive arterial involvement. Remained steady with no new symptoms for three years, then mass had grown around ureters necessitating stents. Histiocytes constantly blocked them necessitating placement of new stents six times in nine months. Had a bilateral nephrostomy in 2003 and it continues to manage the situation, although kidneys are filling up with histiocytes and there is concern over how much longer nephrostomy will be viable. Constant problems with leaking at the port on one side of nephrostomy and constant urine scald/cellulitis on that side. Had one round of experimental chemo in 2000 but it had no effect on mass. Continue to have aching in lower spine, weight bearing bones and more several pain in feet, which tends to come in cycles. Control the pain with Tylenol w/codeine and, on days when I know I'll be on my feet a lot, I take Hydrocodone. Occasional low grade fevers that go away with one dose of Tylenol. Biggest problem is infection in/around leaking port. Tend to have about one day a week that is a "down" day - aching and malaise. Started having problems with one eye in December 2006 that was diagnosed as blepharitis. Problems continued through 2007, with heavy watering. Noticeable bulging in early 2008 and CT showed orbit was filled with mass. Vision is not impaired as of yet. Started Interferon Alpha, 3MIU 3x/wk, 3/31/08. CT after three months show no change, good or bad, in orbit. No problems with Interferon as long as I get plenty of sleep at night.&n bsp;	Masses in abdomen, kidneys, lower spine, hips, legs, knees, feet, orbit and pituitary
16	M	1950	June-05			Bone loss in lower jaw. (waiting for rest of information from patient)	
17	M					(waiting on information from patient)	

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Case #	S e x	YOB	Treatments Tried	Current Treatment	Current Status	Last Update Date
10	F	1967	Initial treatment was Vinblastine 1 x week, Aredia once every 3 weeks both for 7 months. Naxin (pain med) 2 x day, Calcium w/D for 3 months. Symptoms were improved. Prednisone for 8 months beginning at 100 mg/day stepping down to 0 mg.; Interferon as high as 9 mil x 5/week in 2008 to stabilize the disease. Had LPI 3/08 to decrease head internal pressure.  In 2010 Interferon (9 mil) 2 x week; Vinblastine once every 3 weeks; Aredia 1 x month, Naxin 1 x day. Infusion of pamidronate every 30 days. Shiatzu and acupuncture.	For last 6 months treated with 1X9mil + 1X6mil per week . Week ago reduced the dose to 2X6mil per week as result of bad quality of life . Uramux - 750mg per day to resolve internal head and eye pressure . Vinblastin - 1X60ml once in 8 weeks Aredia - once in a month Minirin spray - sniffing 1-2 times per day Nerocin - for pain	Stable, but not feeling well.	12/2/2010
11	M	1952	Three rounds of chemo, vinblastin, 2cda, didn't help. 2 radio active injections of Samarium 265 (or 285) didn't help. On morphine for 33 months. highest level 400mg per day, off in 11/2001 with the help of brain wave stimulation.	Pamidronate every 60 days to prevent bone break down.	Osteoprosis in my lower back.	7/13/2008
12	M	1940	Round 1- 24/7 of 2cda chemo; 7/04 Round 2 was 1 treatment each for 4 days; 9/04 Round 3 was 1 treatment a week for 6 weeks.	Prednisone 10 MG per Day	No new Masses since 09/02; lots of leg pain, Eyes, Leg Weakness. Lower back or left hip has pain along with legs. But patient is glad to be up and around	2/15/2009
13	F	1955	For Eyes: Cortisone; Radiography; Imuran Retroperitoneal growths: Tamoxifen.	ECD meds are combination of Imuran (for eyes) and Tamoxifen (for retro- peritoneal growths). Other drugs at present: Oroxine: for thyroid; Amaryl / Diaformin: for diabetes; Cartia / : for coronary artery disease; Lipitor: for cholesterol control; Atenolol & Karvezide for blood pressure; Panadol if needed for pain relief (avoided when possible); Tea Green for removal of radicals/ toxins; Fish Oil for arthritis	Condition remains stable (2009) but no indications the ECD growths are reducing. Lots of walking to maintain mobility. Pilates & Osteopathy to maintain flexibility. Muscle spasms / strains / pain happens frequently and easily. Bone and muscle pain is present continuously but at variable levels of intensity. The use of a Homedics Percussion Massager (model PA-MH) reduces incidence of muscle cramps, especially those occurring during sleep	Most recent doctor's visit: Feb. 2009. Most recent update of this info: 2/2009
14	M	1964	Chemo - vp16 topicide - 3 days of treatment in a row each month (did for 3 years, stopped for summer of 2008 due to side effects)	DDAVP (for diabetis insiudus); Synthroid; Prevacid; Lopressor; Lasix; Lovastatin; Humalog; Nutrophin; Testosterone injection; Lyrica; pling to start interferon on Oct 13, 2008	No new tumors, old ones are dying; Hydrotcort; worried about losing eyesight	9/20/2008
15	F	1950	One round of chemo, otherwise just treating symptoms	Interferon Alpha 3MIU 3x/wk since 3/31/08, pain medicine as needed, Prednisone, 10mg, daily, BP medicine needed as a result of Prednisone	On disability as of 5/31/2003, able to continue with all normal activities except for walking distances. Self care for nephros-tomy which sometimes needs bandage changes as often as every two hours. CT shows no change 3 mos. after starting interferon treatment. Very low energy, hip/leg/foot pain and loss of flexibility in lower extremities.	7/12/2008
16	M	1950				
17	M					

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18	F	1932	Spring 2008	74	76	First symptom was double vision. After repeated attempts to correct the problem with glasses it became obvious something might be really wrong. August 2008: Scans showed no change in eye, abdomen, or mass at base of skull. First skeletal survey so done and bone involvement was seen. The eye has had noticeable improvement in the protrusion, but is not documented by the radiologist as the initial scan in fall '07 and the more recent one in Feb08 were done at two different locations.	bilateral. xanthogranulomas in her eyes; a mass in her abdomen; a mass at the base of her skull in the spinal canal; showing areas on her proximal tibias
19	M						
24	M	1945	2007 suspected; confirmed Jan. 2008	62	62	<ul style="list-style-type: none"> <li>• In January 2007, patient complained of right ankle ailment: slightly swelling at time; still remnants of "black and blue" from a softball injury sustained four to five years earlier when stepping on a base and catching on back edge with full weight. This led to a comprehensive battery of tests and exams (metabolics; chest and pelvic x-rays; Thoracic, abdominal and pelvic CT scans; and a venous extremity with doppler of the right leg. All results were normal - except the CT scan of the pelvic/abdominal region indicating two soft tissue masses surrounding both kidneys.</li> <li>• Other than discovery of the two tissue masses, no other symptoms experienced. A succession of five medical specialists (hematology, oncology, radiology, urology, and nephrology) at two major hospitals and two biopsies resulted in a preliminary diagnosis of retroperitoneal fibrosis (RPF). Two biopsies (1st - local left renal; 2nd - laparoscopic left renal) were conducted in succession over two months; each proved inconclusive. A review of CT scans and xrays in pelvic/abdominal area reveals NO soft tissue around the aorta in this region (clinical cases define RPF as having such soft tissue in this region. A bone scan with TI-99 led to a change in diagnosis to Erdheim-Chester disease in September 2007. Patient during this period was frequently asked by medical professionals how he felt. He always responded "Great! It's only you doctors who tell me something is amiss."</li> <li>• Other than the two tissue masses surrounding the kidneys and the "long bone evidence" in both legs and arms, there is no other involvement of any other organs, to include the brain, eyes, lungs, etc. These organs are CT scanned periodically (every 9-12 months) monitoring any change. Metabolically, all organs were performing within their norms and these are checked monthly.</li> <li>If I could offer any encouragement to others, it would be these two adages: "Dream/Plan as if you will live forever; live as if you will die tomorrow." and "Forget the potholes in the road of life. Celebrate the journey."</li> <li>• Be strong! Keep the faith!</li> </ul>	Kidney, bone
31	F	1979	Summer 2007	27	27		Long bones, heart, brain, pituitary gland, and my optic nerves were crushed causing me to lose my sight.
32	F	1938	June-05	55	65	Liver enzymes elevated; red spots on skin; elevated sed.rate (100); chest x-ray showed unusual spots in lungs; sinus problems. Occasional joint pain, fatigue, anemia, CT scan showed unusual material in abdominal area, vision problems, thyroid deficiency. Lung biopsy at Mayo in 2003 showed ECD. Consulting doctor knew nothing about ECD.	Lung, eyes, abdominal, thyroid
34	M	1968	January-09	39	41	Leg, knee & ankle swelling, joint pain, sleeplessness, weight loss (this was prior to treatment, symptoms have improved some now)	Legs, kidney, heart, lungs
35	M	1958	April-09	49	50	Initially: fatigue, swollen ankles, night sweats, and high blood pressure. Currently: fatigue; pain in shins, thighs, and upper arms; fevers; night sweats; headaches; muscle weakness	Kidneys (both surgically removed and subsequent live-donor transplant) Spinal cord (debulking surgery to remove growths from thoracic spine) Eyes (growths on both optic nerves -- the right more pronounced than the left. No additional growth in the past 8 months)

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18	F	1932	Dec 2007: Tried 1 month steroid burst which showed mild improvement but with severe side effects. June 2008: Began interferon at 6mil. Aug 2008: Interferon lowered to 3mil due to adverse side effects. Sept. 2008: Again reduced the interferon dosage down to 1 MIL 3Xweek due to side effects of exhaustion, malaise & weight loss	Interferon 1MIL 3xweek	Deceased 2010	6/20/2010
19	M		Interferon		Deceased 2009	
24	M	1945	Prednesone was utilized for the first four months in concert with CellCept. Starting month: 40 mg/day. Second month: 30 mg/day. Third month: 20 mg/day. Fourth and last month: 10 mg/day. CellCept was taken from the first day/first month at the dosage of 1000 mg, twice a day and continues.	CellCept 1000 mg x 2 per day.	No visible symptoms. Fairly healthy and physically active throughout life; accomplished many goals and still has a few more to tackle	9/29/2010
31	F	1979	Radiation, cladribine, interferon	None	I am in remission. I use a blind cane to get around.	10/10/2010
32	F	1938	None	None	Am concerned about anemia, lungs, eyes, plus whatever may be a problem in the future that is not currently affecting things	2/21/2009
34	M	1968	No other treatments tried	180 mcg of Pegasys weekly injection. Was at 150 mcg for 18 months, but have seen slight enlargement in lymph nodes so Dr increased injection dosage to see if this helps. Will see dr in October 2010 to see if there is improvement. No other sites increased in size. If there is no improvement Dr. will try Gleevec.	Currently I am blessed, still able to work fulltime & do everything I did before. I do have constant leg pain, but nothing I can't tolerate. Take Tylenol when needed. I have lost a great amount of weight as a side effect from the pegasys/interferon. Am nervous about the possibility of taking Gleevec.	8/15/2010
35	M	1958	When first diagnosed, doctors believed (or hoped!) that the Rx'd immuno-suppressant & Prednisone might have a positive effect on the ECD. However, only 2 months after the transplant his spinal cord was attacked, again by ECD growths. Then for a year no new growths made their presence known. Personal MD communicated with Dr. Vaglio in Parma, Italy, and the Prograf was switched to Rapamune. The Rapamune is an immuno-suppressant that (hopefully) will also have anti-proliferative properties. An incidental finding on a head CT revealed the growth on the optic nerves; he retains full field of vision in both eyes. In a repeat CT 8 months later, no further growths on the optic nerves. We are encouraged that the Rapamune is working. :)	Prednisone 10 mg/day (originally prescribed as part of the transplant immuno-suppressant) Rapamune 3 mg/day (immuno-suppressant for kidney transplant, as well as potential anti-proliferative properties) For pain: Oxycodone & Fentanyl patches	Walks unaided, although has transient spells of weakness that causes him to hold onto something stable. Feeling OK some days; weakest at beginning of the morning then usually improves as the day progresses. On a scale of 1-10, pain is often 8.5 - 9; however, pain meds make it possible to be productive.	8/15/2010



# ECD Case Log

Case #	Sex	YOB	Dx date	Age at Sx onset	Age at ECD dx	Symptoms	ECD Involvement
36	M	1953	March-99	43		First sign was double vision, but the neurologist couldn't find the cause. In 1998 more problems with eyes. Vision was not stable and left eye was bulging. Diagnosis was given as Graves disorder. A CT-orbita showed tumors behind both eye-balls in the orbita. In January 1999 a biopsy was taken, a lipid deposit called xanthogranulomas was found. A lot of scans (thorax, abdominal, skeleton, hollow pipe-bones, MRI, SMS), biopsy of a kidney, bone biopsy, bone marrow biopsy and tests (blood, thirst) followed and in March 1999 a team of internal medicine doctors were convinced of ECD diagnosis with a beginning diabetes insipidus. The disease had spread out in my body: xanthogranulomas is behind my eye-balls, spots of lipid deposits are found in the bones (hollow pipebones above knees, in hipbones, shoulders, chest, skull, lungs, around the kidneys, around the aorta and cerebral membrane (along the tentorium). In an attempt to save the optic nerve, Cytostatica Solu-Medrol iv followed by another cytostatica called Endoxan iv were tried with no sustainable effect. June 1999 a decompression of the orbita was tried where bones in eyesockets and cavity of nose were removed to make more space in the orbita with no sustainable effect. 18 sessions of Radiotherapy with a temp. improvement in vision. Jan. 2000 a „debulking“ surgery (removing lipid deposits out of the orbita behind left eyeball) was successful and sight was regained in left eye. I have learned that it is important to believe in the good things of life, enjoy each day and make each day a good one with my wife and kids, my family and friends. I discovered my sources of energy and fun, and they brought new and unexpected dimensions, which surprised me so much. For example, I found my guitar again. Making songs and music give me power, satisfaction and a lot of fun.	Eyes, bones, lungs, kidneys, heart, brain, chest, pituitary gland
37	M	1938	June-05	61	61	Edema, Shortness of breath and on oxygen. Tired all the time.....Always aching.	Left Plueral Area
38	F		Approx. 2006			3 years of bi-yearly follow up with bone gammagraphy she was found to have more lesions in the ribs, liver and spine.	Ribs, liver, spine
40	M	1955	Jan. 2003, at death	45	48	January 2000 developed Pleuritis. After nine weeks of rest he got sick again with Pneumonia. Recovered, but had trouble breathing normally when exercising. Test showed lung capacity was decreased to 55%. Underwent multiple blood tests and biopsies. No diagnosis means no treatment. Physical condition slowly decreased and treated with antibiotics or prednisone. In July 2002 underwent an open lung biopsy. Biopsy showed no cancer or asbestos. January 2003 was given one gram of Prednisone treatment.	Lungs, heart, brain
44	M	1965	February-09	43	birth? Or 43	Knee pain since I was 17. Poor balance and occasional slurry speech at age 43. Low kidney function, Higher level of thirst since I was 26.	Balance and speech, kidney function, and teeth
52	F	1948	March-03	54	51	Loss of alveolar bone (supports teeth) in mandible (lower jaw) - began in 1999. Have lost 5 posterior teeth in lower jaw since 2001; occasional leg pain since 2000; hole in left hip DX in 2002; bulbous eyes, headaches, swollen optic nerves (10-08).	bones, lower jaw, behind eyes
53	F	1955	July-09	51, possibly earlier	53	Symptoms first began as swelling in legs, fatigue/no energy, severe cough, shortness of breath, feeling of fullness in chest, bruising and pain of the legs, changes in breasts. In July 2009 after several doctors could not diagnose and symptoms had gotten much worse, we went to Mayo Clinic where 1700 cc's of fluid were drained from the pericardium of the heart after discovery via echocardiogram. Fluid continued to be drained every 4-6 hours via permanent drain. Other symptoms had developed such as back pain, tingling in extremities, pain in jaw, fluttering of peripheral vision, some difficulty sleeping. Diagnosis was made via biopsy of mass surrounding kidneys (retroperitoneum) that was discovered. A pericardial window was placed on July 14, 2009, which fairly quickly stopped the fluid buildup and alleviated the severe chest discomfort.	Retroperitoneum, both eyes, jaw, ends of leg bones, shoulders, sternum/chest cavity, pericardium with some infiltration into heart.
55	M	1961	August-09	45	48	2004 – Xanthalamus on eyes, high cholesterol (may be unrelated to ECD) 2006 – slight diabetes insipidus (increasing in subsequent years) then stabilizing in March 2010. 2007 – lowered testosterone levels, mood swings, low libido etc. 2009 – leg aches, discovered increased prolactin levels.	Slightly damaged pituitary gland confirmed by scans in July 2009. Xanthalamus in eyes shown to contain some Touton cells related to ECD. Surgery in Dec 2009 to remove yellow / Xanthalamus patches was successful, Xanthalamus have not returned as of Aug 2010. Classic ECD scan when bone scan completed in July 2009.

ECD Case Log

Case #	Sex	YOB	Treatments Tried	Current Treatment	Current Status	Last Update Date
36	M	1953	Drug cocktails over the past 10 years have included: prednison-cyclosporine-octreotide-minirin-renitec-fosamax prednison-endoxan/cyclofosamide-minirin-renitec-fosamax prednison-diflucan-minirin-renitec-fosamax prednison-endoxan-enbrel-octagam/gammaglobuline-minirin-renitec-fosamax-losec prednison-endoxan-octagam-minirin-renitec-fosamax In 2001-2002 an experimental treatment with Lutetium 77 (an internal radiotherapy was tried.)	10 mg prednison/day 100 mg azathioprine/day 0.1 minirin/day 30 mg APD iv/ 3 months 5 mg enalapril maleaat/day 1000 mg calcium/day	Stable. No pain, but energy has limits, yet "energy enough to do enjoyable things.	2/18/2009
37	M	1938	Prednisone....Somewhat helpful	Prednisone 10 mgs twice a day	On oxygen, pain level of 7, using walker to get around	8/15/2010
38	F		None	None	Experiencing bone pain.	2/23/2009
40	M	1955	Antibiotics, prednisone	n/a	Deceased	2/4/2009
44	M	1965	Cladribine	Gleevec 400mg (Improved energy, no current pain)	working full time as a Social Worker, still driving. Living a normal, but altered life. Since on Gleevec there has been a slight improvement of the disease and an increase in energy with only slight GI side effects.	8/15/2010
52	F	1948	In 2003, I took Fosamax (1/wk) for 5 yrs - discontinued 2 yrs ago. Surgery 8/09 for decompression of left eye; 9/09 for decompression of right eye. 5 MIU Interferon alpha-2b injections 3X/wk since 12/09 Nsaid as needed for pain	5 MIU Interferon alpha-2b injections 3X/wk since 12/09 Nsaid as needed for pain	Continue to experience occasional leg pain; unexplained, occasional joint pain ; double vision in evening if watching TV; fatigue from Interferon. I still work as a psychotherapist part time and work on remaining positive, which I believe has helped me stay relatively asymptomatic.	8/17/2010
53	F	1955	Stronger narcotic pain meds had negative side effects.	Currently, only taking natural supplements and vitamins under the direction of a homeopathic physician, and Ibuprofen daily for pain. Went through 6 rounds of Cladribine chemotherapy as prescribed by Mayo Clinic which appeared to have significantly shrunk the masses in the chest/heart, and retroperitoneum, less improvement behind eyes and other areas but no worsening. Now take Ativan for sleep, and Thyroid medication for many years. Will have new sets of scans done soon. Latest echocardiogram showed heart/fluid is stable, actually improved since last echo 6 months ago.	Still have fatigue, needing 10 hours of sleep per night, plus frequent naps. Leg pain continues, some days worse than others. Also, arm pain and sensitivity/tingling. Difficulty walking or standing for very long. Overall weakness, loss of strength. Residual bruising of lower legs, minimal swelling. Sensitivity to heat. Skin changes: small white raised "bumps" on face, arms. Occasional cognitive difficulty with memory, organization skills.	8/24/2010
55	M	1961	A healthy diet avoiding saturated fats, high sugar and high meat intake. I eat fish regularly and lots of vegetables. I am not doing enough exercise currently. I am taking various herbal / naturopath recommended items for general health: a) Turmeric extracts BCM-95 for anti-inflammatory effect b) "adpatogen" herbal treatment for adrenal stimulation. (liquorice and other herbs) c) Fish oil (2 x 1000 mg pills per day) with EPA:DHA ratio. d) Specialised Multi-Vitamin pills to give daily intake of a. Vitamin A – 15,000 IU; b. Vitamin C - 850 mg c. Vitamin D3 – 1000 IU; d. Vitamin E – 400 IU e. Thiamine – 40 mg; f. Riboflavin – 11 mg g. Niacin – 160 mg; h. Vitamin B6 – 10 mg i. Folate- 100 mcg; j. Vitamin B12 – 450 mcg k. Biotin – 400 mcg; l. Pantothenic acid – 413 mg m. Calcium – 300 mg; n. Iodine – 225 mcg o. Magnesium – 285 mg; p. Zinc – 15 mg q. Selenium – 200 mcg; r. Manganese – 6 mg s. Chromium – 20 mcg; t. Molybdenum – 100 mcg u. Potassium – 90 mg; v. Boron – 3 mg w. Choline Citrate - 100 mg; x. Vanadium – 100 mcg	1) Daily 50 mg / day Testogel (testosterone supplements) 2) ½ pill (0.5 mg each) twice a week of Cabaser (cabergoline) to reduce prolactin. 3) Daily ½ pill (100 µg) of Minirin (desmopressin) to supplement lack of anti-diuretic hormone. Reduce effects of DI.	I feel good apart from slight leg aches. My well-being is so much better since the testosterone / ADH treatment stated in early 2010. Jan. 2011 Update: 1) Perhaps unrelated to ECD, but I have cured my asthma in the last 6 months by rigorous avoidance of preservatives (the main culprits being soft drinks ("pop") and cider. 2) My eye xanthelasma have disappeared which I believe strongly is due to avoidance of dairy fats (and replacement with oil fish and nuts- almond etc., and no peanuts)	1/16/2011

# ECD Case Log

Case #	S e x	YOB	Dx date	Age at Sx onset	Age at ECD dx	Symptoms	ECD Involvement
60	M	1971	Fall 2009	36	38	Fatigue, nausea, abdominal pain, bone pain, headaches, frequent urination	retro-peritoneal tumor that presses on kidneys/ureters, femurs, hips, lumbar and cervical vertebrae, sinuses
65	M	1948?	June-05	33	57	<p>1981, age 33, I was dx with Diabetus Inseptus cause unk. Went on DDAVP.;</p> <p>Late eighties passed out at church, went to hospital, checked out ok.</p> <p>1999 Had cateract surgery on right eye, Had fluid in heart sack had to be removed.</p> <p>2000 had cateract removed from left eye.</p> <p>2001 went to the V A Hospital to be checked for effects of agent orange, C T scan showed what they called fatty tissue around my liver. nothing done.</p> <p>1995 Dx with type II diabetus, treated with pills under control</p> <p>2000 started haveing trouble breathing (short of breath all the time). My wife commented several times about my breathing.</p> <p>2004 Got sick had pain and discomfort in stomach, thought it was my galbladder, went to the VA emergency room, they said I didn't have classic symptoms, gave me some pain pills, and sent me home.</p> <p>April, 2004, a week later same problem returned, went back to E R. did a C T scan said I had kidney stone,</p> <p>July 2004, I woke up with very dry mouth and a temp. of 104. Went back to E R . They did another C T scan saw the fatty tissue and admitted me. For the Next three weeks they did many test, C T, Blood work, skeletal x-ray, pet scan, heart test, lungs, GI track testing.</p> <p>The end of July 2004, they took out my gallbladder, they were supposed to do laporscopic surgery. When I woke up I had a 12 inch incission. They told me they could not get it out with the small surgery, and hade to do major. More tests followed.</p> <p>Dec 2004 they took samples of the tissue that was growing inside of me.</p> <p>In June of 2005 sent samples to Ohio State University for second opotion. O S U made a dx of ECD.</p>	
77	M	1967	2010	40	42	weakness, loss of strength, balance issues, unsteady gait, slurred speech, tremors in hands/arms, diabetes insipidus, retro-peritoneal fibrosis	brain, pituitary gland, kidneys, long bones

# ECD Case Log

Case #	Sex	YOB	Treatments Tried	Current Treatment	Current Status	Last Update Date
60	M	1971	Prednisone, 40 mg./day that tapered off after 6 months. Felt energetic on it, but had terrible withdrawal from it for 2 months. Interferon, 3 injections/week for 5 months during which tumor grew and disease spread to cervical vertebrae. Felt okay on Interferon other than flu-like symptoms the day after injections.	Started vinblastine on 8/17/10. Initially treated once a week, but had to cut back on the frequency due to a low white blood count. Stents for the kidney were placed on 10/1/10. Also taking oxycodone and lorazepam for pain as needed.	A tumor has caused the left kidney to fold in half and the kidney is failing. The right kidney is functioning, but the tumor is beginning to press on it causing it to lose function. Always tired, frequent abdominal pain, occasional bone pain. Some days are good, some days are bad. Status can change in a matter of minutes.	9/29/2010
65	M	1948?		In 1981 began DDVAP. In Nov 2005 started on interferon, been on it since.	All symptoms have remained, as well as extreme fatigue, shortness of breath, A strong feeling of weakness, no strength, or ambition.	10/19/2010
77	M	1967	prednisone tried for retroperitoneal fibrosis (before ECD diagnosis) and no changes seen. Tapering off prednisone in early 2011.	Started Gleevec January 2011. Also, desmopressin for diabetes insipidus.	Tolerating Gleevec well, but all issues still present. 2nd PET scan in 2 months. Battling swelling from Gleevec. Currently in rehab center recovering from 2 month long hospital stays (Oct 2010 and Dec 2010/Jan2011). Walking with assistance and walker. Hoping to come home soon.	2/12/2011