What is ECD?

- A rare multi-system, non-Langerhans Cell histiocytosis of unknown cause that usually affects adults.
- Characterized by excessive production and accumulation of histiocytes (cells which normally fight infections) within multiple tissues and organs. As a result the tissue becomes thickened, dense and fibrotic.
- Involvement may include long bones, skin, tissues behind the eyeballs, lungs, brain, pituitary gland, kidney, retroperitoneum, pericardium and more rarely other organs. Each patient can have a different combination of organs attacked.
- Unless successful treatment is found, organ failure can result.
- There have been a very limited number of published cases of ECD in the world since it was first described in the literature in 1930 by the American pathologist, William Chester. Most published articles on ECD are anecdotal in nature as the disease is so rare, studies on groups of patients is extremely difficult.

This material was compiled by a group of non-medical people who are trying to raise awareness of Erdheim-Chester Disease. The material in this publication is meant for awareness purposes only, not treatment purposes. Please send any comments or corrections to support@erdheim-chester.org.

In Loving Memory of
F. Gary Brewer, Col. USAF, Ed.D.

and

In Honor of
All Those Who Suffer from ECD

There are many questions surrounding ECD.
The work of the ECD Global Alliance can be found at www.erdheim-chester.org.

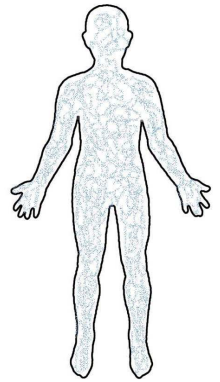
Please contact us with questions or for more information about ECD. Researchers interested in investigating the cause or treatment of ECD are encouraged to contact the organization about possible grant opportunities.

To donate, please send checks to:
The ECD Global Alliance
6375 Thomas Jefferson Hwy
Charlotte Courthouse, Va 23923 USA

The ECD Global Alliance
A Group of Patients and Their Loved Ones

Erdheim-Chester Disease

A rare multi-system non-Langerhans Cell histiocytic syndrome of unknown cause



What is ECD?
What are some symptoms of ECD?
What are some signs of ECD?
How is ECD diagnosed?
How is ECD treated?

What are some symptoms of ECD?

- Varied, depends on organ(s) involved
- Some more common symptoms may include:
  - Bilateral bone pain in legs and knees
  - General symptoms of weight loss; fever; night sweats; muscle and joint aches; feeling of discomfort, weakness, and fatigue (malaise); flu-like symptoms that linger or continue to return
  - Excessive thirst and urination (diabetes insipidus)
  - Balance issues, difficulty walking (ataxia), slurred speech (dysarthria), involuntary, rapid eye movements (nystagmus)
  - Lower back, flank or abdominal pain, often associated with kidney issues (retroperitoneal fibrosis); reduced kidney function
  - Bulging of the eye (exophthalmos) and/or vision issues
  - Sore or bump under the skin (xanthomas), rash
  - Shortness of breath (dyspnea)

ECD affects different organs in different people. As a result, each person will have a different combination of symptoms. This is partly what makes ECD so difficult to diagnose. By taking a systemic view of symptoms it may be possible to test for and diagnose ECD earlier. This will potentially give patients the best chance for a successful treatment plan.

What are some signs of ECD?

Signs are the result of histiocytic infiltration of various tissues. Depending on organ involvement, some of the following signs may be found:

- Bilateral symmetric medullary sclerosis with cortical thickening and coarsened trabecular pattern of the long tubular bones of the extremities
- Abnormal lipid metabolism
- Moderate anemia
- Increased C-reactive protein and erythorocyte sedimentation rates
- High creatinine
- Interstitial lung disease involving accumulations of histiocytic cells and fibrosis in a predominantly perilymphangitic and subpleural pattern
- Soft tissue masses and/or lesions
- Pericarditis, "Coated Aorta" or other cardiovascular abnormalities

How is ECD diagnosed?

- It is often difficult to diagnose ECD
- Bone biopsy typically show sclerotic bone
- Tissue biopsy contain clusters of lipid-laden, foamy histiocytes with signs of chronic inflammation and Touton type giant cells, fibrosis and possible fat necrosis
- Histiocytes express CD 68 positive, CD1a negative, with S-100 protein expression variable and without Birbeck granules

How is ECD treated?

Because of the rarity of this disease, clinical trials have not been conducted. Therapeutic options based on anecdotal experience include:

- Immunotherapy (interferon)
- Systemic corticosteroids
- Chemotherapy (cladribine, vinblastin, vincristine, cyclophosphamide, doxorubicin)
- Immunosuppressants (e.g., Imuran, Cellcept, Sirolimus)
- Imatinib
- Tamoxifen
- Anakinra (trade name Kineret)
- Surgical debulking
- Radiation treatment
- Combination treatment of Remicaid, Methotrexate, and Cellcept

The efficacy of treatments is difficult to evaluate and the disease can be relentless in its course. In general, the clinical course of patients with this disease is variable.

It is important to know there are patients who are living high quality lives with ECD for decades. Because ECD is so rare and publications so few, this information may not be readily available to patients or physicians.