

## What is ECD?

- A very rare disease that can affect many different organs.
- It is not categorized as a cancer, infection or auto-immune disease.
- The cause is unknown, but it usually affects adults.
- It has not been found to be contagious.
- Characterized by excessive production and accumulation of specific cells which normally fight infections (called histiocytes) within multiple tissues and organs.
- Involvement may include leg and arm bones, skin, tissues behind the eyeballs, lungs, brain, pituitary gland, kidney, abdominal cavity, the membrane surrounding the heart, and more rarely other organs. Each patient can have a different combination of organs attacked.
- Unless successful treatment is found, organ failure can result.

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](mailto:support@erdheim-chester.org).

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](http://www.erdheim-chester.org).

Please contact them with questions or for grant information if you are interested in doing research on this disease.

## A Patient's Perspective

# Erdheim-Chester Disease

A rare multi-system histiocytic syndrome of unknown cause



**What is ECD?**  
**What are some symptoms of ECD?**  
**What tests are used to diagnose ECD?**  
**How is ECD treated?**  
**What is it like to have ECD?**  
**What can you do to help?**

## What are some symptoms of ECD?

- Varied, depends on organ(s) involved
- Some more common symptoms may include:
  - Bone pain in legs and knees, usually on both sides
  - General symptoms of weight loss; fever; night sweats; muscle and joint aches; feeling of discomfort, weakness; fatigue; flu-like symptoms that linger or continue to return; and many others
  - Excessive thirst and urination (diabetes insipidus)
  - Balance issues, difficulty walking, slurred speech, involuntary, rapid eye movements
  - Lower back, flank or abdominal pain, often associated with kidney issues; reduced kidney function
  - Bulging of the eye
  - Sore or bump under the skin, rash
  - Shortness of breath
  - Heart issues

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 tests are used to diagnose ECD?

- It is often difficult to diagnose ECD and may take years to diagnose
- Bone biopsy, tissue biopsy, ultra sounds, bone scans, PET scans, CT scans and MRI scans are all often used to diagnose and monitor the disease
- Some believe this disease is under diagnosed

## How is ECD treated?

Because of the rarity of this disease, there is no treatment plan approved by the medical profession as a whole. Based on individual experiences, the following treatment plans have been used with varying degrees of success:

- Drugs such as immunotherapy, steroids, chemotherapy and other treatments found to help in the fight against certain cancers
- Surgery to remove tumors and parts of tumors
- Radiation treatment

It is difficult to evaluate how well any of these treatments work. The disease can be relentless in its course. In general, the prognosis for patients with this disease is variable. It is important to know there are patients who are living high quality lives with ECD for decades after diagnosis. Because ECD is so rare and little research has been done on the disease, there is not much information available to patients or physicians.

## What is it like to have ECD?

ECD patients face many challenges. Some of these may include:

- Extremely tired ALL the time
- Pain
- Side effects of many of the treatments are difficult to tolerate
- Feelings of aloneness, frustration, anxiety, fear

Some patients experience these and other challenges for long periods of time. Other patients are able to participate in life for long periods of time with few of these issues.

## What can you do to help?

- Be an advocate. Help educate others about this disease and let your law makers know that funding for rare disease research and support is important to you.
- Be supportive. If you know someone with the disease, help them with daily activities, listen to them and just take time to be with them.
- Be generous. Donate to

The ECD Global Alliance  
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