

# HISTIOCYTOSIS ASSOCIATION™

*A Rare Community*

## Fact Sheet Erdheim-Chester Disease

### What is Erdheim-Chester disease?

Erdheim-Chester disease (ECD) is a rare form of non-Langerhans cell histiocytosis. It involves the excessive production of histiocytes, which are a type of white blood cell. These cells, which normally help fight infection and injury, accumulate in different organs and tissues and can result in a variety of symptoms, including organ failure.

ECD is a disease that most often becomes apparent in middle age, with an average age at onset of 53 years. It can affect both men and women. The rate of occurrence is not known, although it is believed to be under-diagnosed and/or misdiagnosed. At the present time, it is not categorized as a cancer, immune disorder, or infection. It is not believed to be contagious or hereditary. The cause is not known.

This disease mostly affects long bones (arms and legs), but can occur in the tissues behind the eyeballs, kidney, skin, brain, lung, heart, pituitary gland, and a part of the posterior abdominal wall called the retroperitoneum. ECD is sometimes mistaken for Langerhans cell histiocytosis (LCH). However, a biopsy of the affected tissue differs in a number of ways from LCH and can establish a definite diagnosis. The cells in an ECD stain for the same proteins as Juvenile xanthogranuloma (JXG) disease but the clinical presentation and age is different. The symptoms and course of the disease depend on the location and extent of the involvement of the internal organs (i.e. the disease outside the bones).

### What are the symptoms of Erdheim-Chester disease?

Symptoms of ECD may include:

- » Difficulty with coordination, slurred speech, behavior disorders, and rapid, involuntary eye movement
- » Pain in bones (mostly arms and legs)
- » Bulging eyes, difficulty with vision including double vision, yellow bumps on eyelids
- » Kidney pain and dysfunction
- » Lower back and stomach pain

### What is the treatment for Erdheim-Chester disease?

Because this is a very rare disease, no widely-accepted treatment plan has been established. Various treatments, however, have been used by individual doctors with different levels of success. These include systemic corticosteroids which is a kind of drug based on hormones that work to reduce inflammation in the body. Another treatment is immunotherapy, also known as interferon, which restores the ability of the immune system to fight off infections; it is usually given as an injection of chemotherapy, drugs normally given in the vein to control the over-production of histiocytes. Also, radiation and surgery may be used for treatment.

### Histiocytosis Association

The Histiocytosis Association is a global nonprofit organization dedicated to addressing the unique needs of patients and families dealing with the effects of histiocytic disorders while leading the search for a cure. It is the only organization of its kind, connecting the patient and medical communities to:

- » Grow and share knowledge of histiocytic disorders,
- » Provide critical emotional and educational support to patients and families, and
- » Identify and fund key research initiatives that will lead to a world free of histiocytic disorders.

Histiocytic disorders affect fewer than 200,000 people. It is thus considered an “orphan disease” and, as such, does not receive substantial government funding for research. The Association relies on contributions from corporations, foundations and individual donors to fund critical research, build awareness and conduct community outreach initiatives.

### For more information, contact:

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