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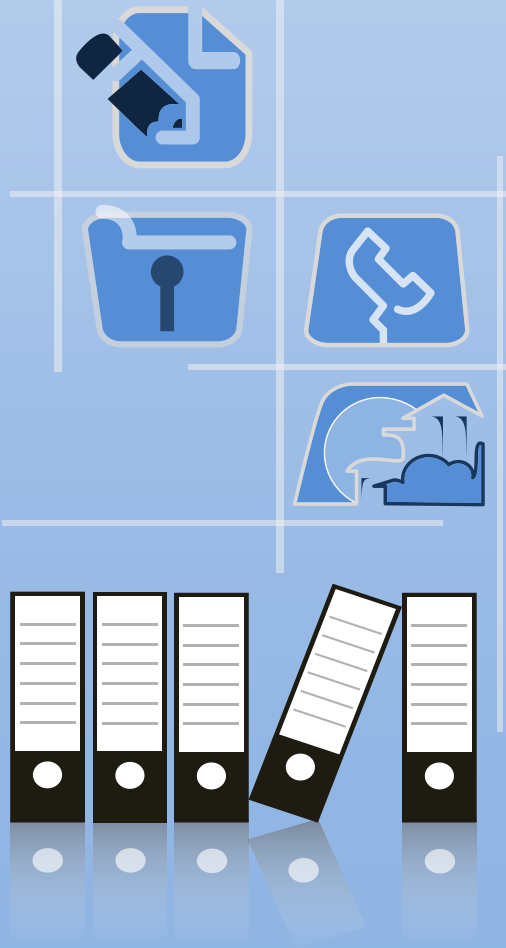
Erdheim Chester

ECD



Histiocytosis UK

Introduction

Despite the misery it causes, Histiocytosis is too rare a disease to have generated substantial research in medical circles. Unfortunately, for every child or adult fighting for his or her life, the pain and suffering are just as severe for children and adults afflicted with other better known disorders receiving funding.

For the children and adults battling these illnesses, there is now reason to hope. To ensure the research continues, we ask for your help, to complete the funding puzzle.

Our awareness and research programmes provide a beacon of hope for the many children and adults battling Histiocytosis, to ensure this research continues we ask you to pledge your support.

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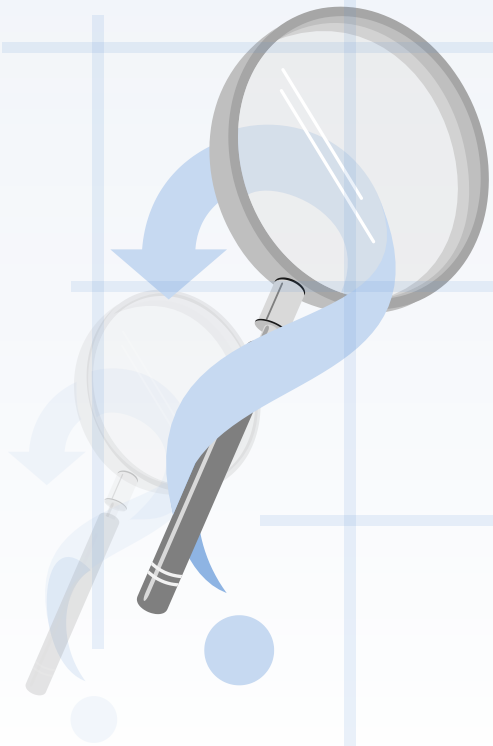

What Is Histiocytosis

What is Histiocytosis?

Histiocytosis is an umbrella term applied to a group of rare diseases, characterised by increased numbers of white blood cells called histiocytes in the blood and tissues. In all forms of histiocytosis, these cells, which are part of the protective immune system, begin to attack the body, targeting many organs of the body including the bone marrow, liver, spleen, lungs, skin, bone and brain.

The prognosis for patients varies greatly depending on the form of histiocytosis.

Please be advised that all the information you read in this document is not a replacement for the advice you will get from your consultant and their team.

Who we are?

Histiocytosis UK is a registered charity dedicated to promoting and funding scientific research into uncovering not only the causes of all histiocytic diseases, which include Langerhans Cell Histiocytosis and Haemophagocytic Lymphohistiocytosis, but also ensuring early diagnosis, effective treatment and a cure.

The Charity aims to support patients and their families by means of information and awareness as well as raise public and professional awareness of histiocytic disorders. Its team of Trustees include the UK's leading paediatric LCH and HLH specialists.

Erdheim Chester Disease
ECD

Erdheim-Chester disease 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, then gather in different organs and tissues and can result in a variety of symptoms, including organ failure.
Erdheim-Chester is a disease that most often becomes apparent in middle age, with an average age at onset of 53 years. It can affect 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 although some cases have the BRAF V600E mutation also found in LCH and cancers such as melanoma and thyroid cancer.

The first two cases of ECD were reported by scientists Jakob Erdheim and William Chester in 1930. In 1972, Dr. Ronald Jaffe reported a third case and coined the name Erdheim-Chester disease (ECD).

This disease mostly affects long bones (arms and legs), but it 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 retro peritoneum. Erdheim-Chester is sometimes mistaken for Langerhans Cell Histiocytosis. However, a biopsy of the affected tissue differs in a number of ways from LCH and can establish a definite diagnosis. The cells in ECD stain for the same proteins as Juvenile Xanthogranuloma (JXG) 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).

Because this is a very rare disease, no large studies have been performed, and no treatment plan has been established that is widely accepted. However, various treatments have been used with limited success. These include steroids, immunotherapy (treatment to restore the ability of the immune system such as interferon), chemotherapy to control the over-production of cells, the use of high-energy rays (radiation therapy), and/or surgery. Some patients with the BRAF V600E have been treated with Vemurafenib, which targets this mutation. While these treatments may control the symptoms and growth of the disease, there is no known “cure.”

Erdheim Chester can be life-threatening with complications such as heart failure, severe damage to the lungs, and kidney failure. However, with treatment, there are patients who are able to live a near-normal life.

FAQ - ECD

Erdheim-Chester Disease

1. What causes Erdheim-Chester Disease (ECD)?

Erdheim Chester disease involves the excessive production of histiocytes, which are a type of white blood cell. What causes over-production of these cells is not yet known, although some cases have the BRAF V600E mutation also found in LCH and cancers such as melanoma and thyroid cancer.

2. Is there a cure for Erdheim-Chester Disease?

The best treatments available today may control and sometimes shrink the tumors associated with the disease. However, we usually don't use the term "cure" for this disease, since no specific amount of time without active disease has been established to determine that a patient is cured.

3. What are the different therapies/treatments commonly used to treat Erdheim-Chester Disease?

To date, there is no universally accepted treatment for Erdheim Chester. Various treatments, however, have been used with variable success. These include steroids, interferon, radiation, surgery, and chemotherapy such as vinblastine, vincristine, Cytoxan (cyclophosphamide), Adriamycin (doxorubicin), and 2CdA (cladribine). Other drugs have also been including Vemurafenib, which targets the BRAF V600E mutation.

4. Can an infant be tested at birth for ECD?

No, a biopsy of the affected tissue, rather than a blood test, is required for diagnosis and unless the patient has a lesion this could not be performed.

Possible Side Effects

What are the possible side effects of interferon?

More common signs/symptoms include:

a. Flu-like symptoms (Fever, chills, headache, dizziness, fatigue, muscle aches, nausea, vomiting, diarrhea)

b. Irritability/depression Decreased appetite

c. Irregular heart rate

d. Decreased blood counts (red cells, white cells, and clotting cells)

e. Liver abnormalities

What are the possible side effects of 2-CdA (cladribine /leustatin)?

More common signs/symptoms include:

a. Flu-like symptoms (Fever, chills, headache, fatigue, nausea/vomiting)

b. Decreased appetite

c. Constipation

d. Low blood counts (red cells, white cells, and clotting cells)

e. Skin rash/redness/itching