About the Association

A Rare Community

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
- Identify and fund key research initiatives that will lead to a world free of histiocytic disorders

Partnership for a Cure

The Histiocytosis Association works closely with the Histiocyte Society, an international organization of over 240 physicians and researchers, which is dedicated to studying the histiocytic disorders. Through this partnership, understanding of this disease has greatly increased, and survival rates and quality of life continue to improve.

Community Outreach

While the search for more effective treatments and a cure continues, the Histiocytosis Association is dedicated to supporting and empowering the patients and families who live with these diseases every day.

Funding the Association

Histiocytic disorders are considered "orphan diseases." An orphan disease is one that affects less than 200,000 individuals in the United States.* Subsequently, these disorders do not receive a high priority for government-funded research. The Association relies on contributions from corporations, foundations and individual donors to fund critical research, build awareness and conduct community outreach initiatives.

**Rare Disease Act of 2002*

You are not Alone

While the search for more effective treatments, and ultimately a cure continues, the Histiocytosis Association is dedicated to informing and empowering those who live with histiocytic diseases every day.

Whether newly diagnosed and learning to navigate this rare diagnosis or searching for strength and support while caring for a loved one, you can turn to the Histiocytosis Association to connect you with a community who understands what you are going through.

The Histiocytosis Association invites you to become a part of this our rare community. We encourage you to explore our online resources by visiting our website:

www.histio.org

Making a Donation

All donations are tax deductible (Federal Tax ID # 22-2827069). Visit www.histio.org/donate to join us in the pursuit of a cure.

HISTIOCYTOSIS ASSOCIATION
A Rare Community

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ERDHEIM-CHESTER DISEASE

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What is Erdheim-Chester disease?

Erdheim-Chester disease (ECD) is a rare type of blood disease that belongs to the group called as histiocytic disorders or histiocytosis. ECD was previously considered an inflammatory or autoimmune disease, but was recognized as a blood cancer in 2016 by the World Health Organization. This was due to the discovery of cancer-causing DNA changes (mutations) in BRAF- and other genes in biopsy samples from most patients.

ECD primarily affects adults, and average age at diagnosis is around 50 years. Approximately 1,500 cases of ECD have been reported in the literature as of the publication of this document. The disease-causing cells of ECD (histiocytes) can involve any organ system of the body from head to toe, but most commonly affect the long bones of the legs around the knees. Due to the wide variety of manifestations of ECD often mimicking other diseases, it is believed to be under-diagnosed. While the exact cause of developing ECD is unknown, it is not considered a hereditary or contagious disease. The discovery of BRAF and other mutations has revolutionized the care of patients with ECD, resulting in targeted treatments that have the ability to improve patient survival.

The diagnosis of ECD can be challenging and requires review of the biopsy of a tumor specimen in light of clinical and radiographic findings. The classic diagnostic finding of ECD is abnormal imaging of the bones around the knee on a CT scan, PET scan, MRI, or a technetium bone scan. A biopsy of one of the affected sites from ECD using a needle or through surgery is usually conducted and reviewed by an expert pathologist under the microscope for features suggestive of ECD. In addition, testing for mutations of the BRAF and other genes through special tests such as next generation sequencing (NGS) studies on the biopsy specimen or blood may be undertaken to help aid in the diagnosis or treatment. There are many hospitals, laboratories, or companies that perform NGS, and it is recommended to discuss with your doctor about which one to use for your case.

Symptoms of ECD

- Pain in bones (mostly arms and legs)
- Kidney pain and dysfunction
- Lower back and stomach pain
- Difficulty with coordination, slurred speech, and behavior disorders.
- Mood and memory difficulties
- Pituitary deficincies (excessive thirst and urination, low libido, cold intolerance, weight gain).
- Shortness of breath, slow heart rate, swelling of feet, ankles, and lower legs.
- Bulging eyes, difficulty with vision including double vision, yellow bumps on eyelids

How is ECD treated?

Most patients with ECD will require treatment at the time of diagnosis. In some patients who do not have any symptoms or any critical organ involvement like brain or heart, it is reasonable to pursue a “wait and watch” strategy for some period of time without treatment. In these cases, close monitoring by a specialist is needed, and may include repeated imaging studies like PET scan or CT scan. Patients with ECD should have their treatment planned by a team of health care providers who are experts in the disease.

The treatment approach to ECD has evolved significantly due to discovery of mutations in the BRAF and other genes, leading to the successful use of targeted therapies in most cases. Treatments for ECD can be divided into two categories- targeted and conventional treatments. Patients may also consider enrolling in a clinical trial if available near them.

To learn more about the Association and its Scientific Initiatives, Outreach Efforts and Research Program visit www.histio.org.

HISTIO

The Histiocytosis Association is dedicated to raising awareness about histiocytic disorders, providing educational and emotional support, and funding research leading to better treatments and a cure.

A world free of histiocytic disorders.

www.histio.org