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 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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XANTHOGRANULOMA

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What is Xanthogranuloma?

Xanthogranuloma (XG) is a rare type of histiocytic disorder that mostly occurs in children (average age 2 years), known as juvenile xanthgranuloma (JXG), but can rarely occur in adults as well, in which case it is called adult xanthogranuloma (AXG). The cause of XG is unknown at this time, although it can sometimes be seen in association with conditions named as neurofibromatosis type 1 and juvenile myelomonocytic leukemia. Rarely, XG can be driven by genetic changes (mutations) in the MAPK pathway, including the BRAF and ALK genes. These mutations occur in the tumor cells, and not in the DNA that is transmitted to children. Because XG is so rare, no large studies have been performed, and there is no established, proven treatment for the more complicated cases. It is possible for XG to be systemic, meaning it can have an effect on the whole body, not just one part of it. However, the Histiocytosis Association continues to work closely with an international group of physicians, known as the Histiocyte Society, who are dedicated to studying all of the histiocytic disorders. Through their combined efforts, awareness about XG has increased, and progress has been made in the understanding of this disease.

Diagnosis and Workup

The diagnosis of XG relies on reviewing the biopsy of a tissue lesion under the microscope. Occasionally, the dermatologist may make a presumptive diagnosis based on the appearance of the skin lesions. When a biopsy is performed, a small piece of the tissue is obtained so that it can be viewed under a microscope by a pathologist. If the cells in the tissue have certain characteristics, the diagnosis of XG can be made. XG cells are negative for the presence of proteins called CD1a and CD207, while LCH cells are positive for those markers. Instead XG cells are positive for proteins called CD68 and factor XIIIa. The pathologist may sometimes do additional tests on the tissues including mutational testing to help with arriving at a diagnosis. It is difficult to distinguish between XG and Erdheim-Chester disease under the microscope.

In patients with skin-limited lesions and no systemic symptoms or signs, evaluations should include a complete blood count, liver function tests, and ophthalmologic (eye) examinations.

An ultrasound may be performed to see if the XG is in the abdominal organs such as liver or spleen. In addition, depending on the symptoms other tests such as CT scans, MRI scans, or bone X-rays may be done. Rarely, the physician might order a PET scan to look for various sites of involvement.

Symptoms of XG

Symptoms of XG vary greatly from patient to patient. It is also possible to have disease in a particular location without noticeable symptoms or for the disease to impact the whole body without immediate noticeable symptoms. Possible site involvement include:

- Skin (lesions)
- Eye (lesions)
- Bone Marrow (abnormal blood count)
- Liver (inflammation, abnormal blood liver tests)
- Pituitary gland - causing diabetes insipidus *(characterized by excessive thirst and urination)*

How is XG treated?

Treatment varies according to the extent and severity of disease involvement. Most children with skin-limited lesions do not require treatment and can be observed with serial examinations. Surgical removal may be undertaken for several reasons: to obtain a biopsy for diagnosis, when there is an organ-function problem because of disease, for cosmetic reasons or to remove scar tissue. For the small percentage of individuals who have symptomatic or rapidly growing disease, treatment with chemotherapy or low-dose radiation has been reported, although there is no standard treatment that is agreed upon. With eye involvement, steroids may be applied to the surface of a lesion, injected within the lesion, or taken in pill form. Rarely low dose radiation treatment may be given to the eye to prevent visual loss. XG in adults (AXG) tends to be more complicated and is not known to improve spontaneously without treatment. This may involve surgical removal alone or additional therapy depending on the clinical situation.

Most common treatment regimens for JXG or AXG are derived from LCH, such as cladribine, cytarabine, or vinblastine + prednisone. If there is a presence of a mutation (BRAF, ALK, etc.), specific pills targeting those genes can be utilized.

*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