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

The Road to a Cure

The Histiocytosis Association is among the world's leading financial supporters of scientific research into histiocytic disorders. Each year the Association conducts a comprehensive and rigorous grant proposal process, and with the guidance of experts, identifies the most important and promising research studies to receive funding.

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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PULMONARY LANGERHANS CELL HISTIOCYTOSIS

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What is pulmonary Langerhans cell histiocytosis (PLCH)?

Pulmonary Langerhans cell histiocytosis (PLCH) is an uncommon lung disease which usually affects young to middle aged smokers or former smokers. PLCH causes the formation of multiple lung cysts (air-filled pockets inside the lung tissue), which can progress over time leading to loss of lung function.

What causes PLCH?

While the precise cause of PLCH remains unknown, almost all adults with PLCH (about 95%) have either smoked cigarettes or have been exposed to substantial second-hand smoke, but can be part of multi-system disease. It is believed that cigarette smoke causes activation and accumulation of specific immune cells in the lungs (Langerhans cells) which leads to a cascade of inflammation and injury to the bronchial tubes and lung tissue over time. In addition to cigarette smoke, there may also be a subset of patients where PLCH is caused by genetic mutations. Recently, cancer-causing mutations in the BRAF- and other genes have also been found in cases with single-system pulmonary LCH, suggesting that a large proportion of these may be cancerous. Cells from LCH tumors (lesions) are found to have cancer-causing DNA changes (mutations); these include mutations of the BRAF, MAP2K1, and ARAF genes. Previously thought to be an auto-immune disorder, LCH was classified as a blood cancer in 2008 by the World Health Organization.

How is PLCH diagnosed?

The most important diagnostic test in patients with suspected PLCH is a CT scan of the chest (CAT scan). In many patients, the CT scan may show changes that are highly suggestive of PLCH (the presence of cysts and/or nodules). Some patients may need a lung biopsy or additional testing to confirm a definitive diagnosis. Other tests (blood work, ultrasound of the abdomen, PET scan, MRI brain) may be indicated in some patients to understand the extent of disease and whether there is LCH affecting organs other than the lung.

What is the natural history of PLCH?

The natural history of PLCH is variable. Some patients have an excellent prognosis, while others progress at a rapid rate. In general, PLCH is a progressive disease and leads to a gradual decline in lung function over time. The inability to stop smoking is associated with a worse prognosis, and can accelerate the loss of lung function.

How common is PLCH?

The exact prevalence of PLCH is unknown. Current estimates suggest that there are between 25,000 – 50,000 patients with PLCH worldwide. These numbers are likely an underestimate.

Who treats patients with PLCH?

The care of patients with PLCH is often provided by pulmonologists, although sometimes a multi-disciplinary approach with input from other specialists like a hematologist/oncologist or dermatologist may be necessary, depending on which organs are involved.

What are the symptoms of PLCH?

PLCH affects patients in different ways. Some patients have little or no symptoms at all. The most frequently reported symptoms include shortness of breath on exertion,sinus congestion and dry cough. Some patients experience fatigue, weight loss and sometimes even a low grade fever.

Approximately 15% of patients with PLCH may experience a pneumothorax (lung collapse due to leakage of air around the lungs). The common symptoms of a pneumothorax are sudden onset chest pain and shortness of breath, typically more on one side.

About 10-15% of adults with PLCH will also have symptoms due to disease in organs outside of the chest; for example bone pain or skin rash.

What is the treatment for PLCH?

Stopping smoking (or eliminating second-hand smoke exposure) is key to the management of PLCH. Smoking cessation may lead to disease stabilization, and even regression, in patients with PLCH. Treatment options in patients who continue to progress after successful smoking cessation include consideration of certain chemotherapy medications and other targeted therapies. Other medical therapies that may be used to treat PLCH include therapies for pulmonary hypertension (a condition associated with elevated pressures in the lung circulation) and oxygen for selected patients. Because of the high risk of recurrence, patients with PLCH who develop a pneumothorax should undergo procedures such as pleurodesis (remove of excess fluid from the area between the lungs and chest wall) in order to prevent/reduce the risk of future pneumothoraces.

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

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