HISTIOCYTOSIS ASSOCIATION™
A Rare Community

Fact Sheet

Hemophagocytic Lymphohistiocytosis (HLH)

What is hemophagocytic lymphohistiocytosis?

Hemophagocytic Lymphohistiocytosis (HLH) is a rare disorder of the immune system primarily affecting young infants and children, although it can develop for the first time at any age. According to a large population-based study published in Sweden, it was estimated to occur in 1.2 cases per million.

HLH is often referred to as either the “primary” form which is hereditary, or the “secondary” form associated with infections, viruses, autoimmune diseases and malignancies (or cancers).

What are the symptoms of hemophagocytic lymphohistiocytosis?

Symptoms of HLH may include:

- Persistent fever, often high
- Liver and spleen dysfunction
- Coordination problems
- Sudden blindness
- Enlarged lymph nodes
- Seizures, irritability and fatigue
- Immunological dysfunction
- Skin rash
- Abnormal blood tests
- Weakness of face/eye nerves
- Paralysis and coma (very rare)

What is the difference between the primary and secondary forms of HLH?

The primary form of HLH is also known as familial hemophagocytic lymphohistiocytosis (FHLH or FHL). It occurs when defective genes are inherited from either both parents (autosomal recessive) or from the mother alone. Since 1999, five genes have been identified which correspond with five types of autosomal recessive HLH.

Secondary HLH is often diagnosed in older patients, and there is no family history associated with this form of disease. It is not known to be caused by, but may be associated with vaccinations, viral infections such as Epstein-Barr, cytomegalovirus (CMV), herpes virus and other underlying diseases.

What is the treatment for hemophagocytic lymphohistiocytosis?

Treatment of HLH/FHL can include a combination of chemotherapy, immunotherapy and steroids. Antibiotics and antiviral drugs may also be used. These treatments may be followed by a bone-marrow or stem-cell transplant in patients with persistent or recurring HLH or those with FHL.

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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