

Hemophagocytic lymphohistiocytosis (HLH)

- Hemophagocytic lymphohistiocytosis (HLH) is a disorder of inflammation that was first thought to affect only young infants and children but is increasingly recognized in older children and adults.
- 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). In the familial form of HLH/FHL, defective genes are inherited from both parents (called autosomal recessive). Secondary/reactive HLH is usually diagnosed in older patients with no family history of this disease.
- According to a large, population-based study from Sweden, it was estimated to occur in 1.2 cases per million children, corresponding to 1 in 50,000 births. However, this number is probably an underestimation as physicians appear to recognize HLH more readily now than when this was published decades ago. According to a large Japanese study, around 40% of cases occur in adults.
- HLH occurs when the immune system is triggered (most often by a virus) and overreacts in specific ways that make the patient severely ill. Thus, HLH is best thought of as a problem of proper immune regulation.
- Poor immune regulation may be caused by genetic defects (called familial HLH, or FHL) or it may occur in patients with various infections, rheumatologic diseases, or cancers, for reasons that are not entirely understood (sometimes called secondary HLH).
- Genetic testing is usually recommended for young children or adults with recurrent or unexplained HLH.
- Symptoms of HLH include persistent fever, often high, jaundice, skin rash, and abdominal swelling.
- A wide variety of symptoms related to brain involvement include irritability, seizures, fatigue, low or abnormally increased muscle tone, difficulty with coordination, weakness of face/eye nerves and paralysis and coma (very rare).
- It is sometimes difficult to establish the diagnosis of hemophagocytic lymphohistiocytosis (HLH), and the combination of the physical symptoms and certain laboratory tests are required.
- The criteria for diagnosis include diagnosis of a specific gene defect and/or the presence of at least five of the following eight criteria: prolonged fever, blood cell abnormalities, enlarged spleen, increased triglycerides, increased ferritin, abnormal bone marrow, abnormally high T-cell activation, low or absent NK cell function.

**Learn more about
HLH and help us
spread awareness!**

