What is Haemophagocytic Lymphohistiocytosis?

Haemophagocytic lymphohistiocytosis (HLH) is a rare blood disorder commonly found in young children. It is not a well understood disease and therefore can be difficult to treat.

There are two types of HLH, primary and secondary. The primary form is genetic and may be referred to as Familial HLH if this disease has occurred in previous siblings. Secondary haemophagocytic lymphohistiocytosis, whilst as common as the primary form develops following abnormal activity of the immune system. This can occur following an infection or in an immunocompromised child.

A basic description is: Haem meaning blood. Phagocytic relates to the action of the Histiocyte cell and means to engulf foreign material and remove it. Lymph is the name of fluid within the lymphatic system, which forms part of the immune system.

The human body contains many types of cells which help fight infection. One type of cell is called the histiocyte. Histiocytes are made by the bone marrow and may travel throughout the body in the blood. Their job is to help destroy foreign materials and to fight infections.

People with active HLH have overactive histiocytes, as well as lymphocytes (another kind of infection-fighting cell), both of which are white blood cells that may cause inflammation (swelling, redness, heat, pain, and loss of function). These cells then build up in good tissue and can cause damage to a range of organs, including bone marrow, lymph nodes, liver, spleen, skin membranes surrounding the brain, spinal cord or, more rarely, the brain itself.

Signs and Symptoms may include:

- Fever
- Rash
- Tiredness, pale, and shortness of breath - known as anaemia, due to a low number of red blood cells that carry oxygen around the body
- Easy bruising - due to a low number of platelets, which are needed for the blood to clot, known as thrombocytopenia.
• Some children may experience seizures, ataxia (wobbly, unstable movements), hemiplegia (paralysis of one side of the body) or irritability due to the central nervous system being affected.
• Enlarged spleen (splenomegaly), enlarged liver, jaundice.
• Enlarged lymph nodes
• Failure to thrive, weight loss, loss of appetite, lack of energy.

What tests may be required?
• Blood tests
• A CAT scan or MRI
• Bone Marrow Biopsy, skin biopsy, lymph node biopsy, liver biopsy.
• Genetic testing

Further information about these tests is available. Please ask medical or nursing staff and refer to sections 3 and 4.

How is it treated?

Exactly how the disease will progress is a difficult question for anyone to answer. Your child is likely to receive extensive testing and aggressive treatment with various types of chemotherapy and/or other drugs.

The first aim of treatment is to try and get the disease into remission. Control of the disease, if achieved, is temporary in the primary inherited form. The disease always returns sooner or later if treatment is stopped. Bone marrow transplantation offers the best chances of a cure, but many children are not well enough for this procedure. Also, for some a suitable bone marrow donor cannot be found. In secondary HLH the underlying cause of the disease is identified and, if possible, treated. In some cases remission may be temporary and similar treatment for familial HLH may be required.

Your child’s doctor will tell you what you can expect and discuss your child's symptoms, treatment, progress, treatment options, and outlook.

Over the years cancer treatments have been used in patients with HLH/FHL. Consequently, haematologists and oncologists who treat cancer also treat children with HLH/FHL. Whilst this disease is not a cancer, it is a life-threatening disease.
Further Information

- Nurses, psychologists and social workers at the hospital can assist you
- WCH genetic counselling services can help with family counselling.

References:

The facts about HLH

Lymphohistiocytosis
http://www.emedicine.com/ped/topic745.htm

Please refer to section 7 “Sources of More Information” or check the parent’s library in McGuiness & McDermott children’s clinic for further information.