

A second objective is to treat any infectious agent that triggered the disease, for example, viruses and bacteria. This aims to remove the cause of the excessive inflammation. Unfortunately, this may not be enough to stop the excessive inflammation as it may have spiralled out of control.

Thirdly, the ultimate aim for children with a genetic cause of HLH is to replace the defective cells in the bone marrow with healthy cells from a donor by stem cell transplantation.

Treatment is tailored for individual patients and is guided by the type of HLH your child has, the severity of the symptoms, their age and any other underlying conditions.

Both diagnosis and treatment are performed at specialist treatment centres which have experience in dealing with patients with HLH. In the UK, there are about 20 of these specialist centres and care will be co-ordinated from there. To make things easier, some treatment may be carried out in a local hospital near to home. This is known as shared care.

The prognosis for children with HLH has improved dramatically over the last 20 years and there is now a cure rate of more than 50%. Overall survival depends on the type of HLH and the response to treatment.

What is the Histiocytosis Research Trust?

The Histiocytosis Research Trust was set up as a registered charity in 1991 by Dr Jon Pritchard and Paul Kontoyannis and is dedicated to promoting and funding scientific research into uncovering not only the causes of histiocytosis diseases, but also ensuring early diagnosis, effective treatment and a cure.

The H R Trust also aims to support patients and their families as well as raise public and professional awareness about histiocytic disorders. It has strong links with The Nikolas Symposium, The Artemis Association, The Histiocyte Society, Euro-Histio-Net, and the Histiocytosis Association of America.

How You Can Help

Research into HLH has been very limited due to lack of funds and all our research to date has been the result of donations and fundraising. In order to achieve our objective to find a cure as well provide practical support for both patients and their families, we need to continue raising as much money as we can. To achieve this, we need your help.

By raising money and working together, our chances of securing a cure will be even greater.

HLH is indeed rare but for the few it affects, it causes devastation, fear and sometimes, death. But together we can find a cure.



Haemophagocytic Lymphohistiocytosis

Together we'll find a cure



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Haemophagocytic Lymphohistiocytosis (HLH) is a very rare group of diseases. It can be divided into genetic (inherited) and acquired forms both of which may be triggered initially by infections, often viruses. The frequency of inherited HLH is estimated at 1 child per 200,000 children. In the UK alone, there are approximately 15 new cases each year.

The incidence of acquired HLH is unknown but it is thought to be more common than the genetic type.

HLH is caused by an uncontrolled activation of white blood cells called lymphocytes and histiocytes which stop the body's immune system from working properly. This process can be likened to a very severe form of inflammation that the body is not able to switch off. Unfortunately, the immune system is overwhelmed by this activation and functions poorly, thus leaving the child susceptible to infection.

HLH often looks like a normal response to infection and it can take time to realise that the child's immune system is not functioning properly.

Types of HLH

Genetic Haemophagocytic Lymphohistiocytosis

Genetic HLH is also called primary HLH and may be inherited in one of two ways:

Autosomal recessive – this is where the child has 2 copies of the abnormal gene, one from each parent

X-linked – this is passed to boys from one of the mother's X chromosomes which is abnormal.

There are two main types of genetic HLH. Those called Familial HLH and those that are related to very rare syndromes associated with immune deficiencies such as Chediak-Higashi Syndrome 1 (CHS-1), Griscelli Syndrome 2 (gs-2) and X-linked lymphoproliferative syndrome (XLP).

Familial HLH

About one in every 200,000 children is diagnosed with Familial HLH. Most, around 70-80%, develop symptoms before the age of 1 and a few, approximately 10%, experience symptoms within the first 4 weeks of life. In the same family, children with familial HLH usually develop symptoms around the same age.

Acquired Haemophagocytic Lymphohistiocytosis

Acquired HLH is also called secondary HLH and can occur at any age. The frequency is unknown but it is thought to be more common than the genetic type. Like the genetic types of HLH, acquired HLH is usually triggered by an infection, often a virus. HLH can also occur in children with some cancers.

Macrophage Activation Syndrome

Macrophage Activation Syndrome (MAS) is an extremely rare condition that occurs in both children and adults with auto-immune diseases, such as rheumatoid arthritis. It has the same features as HLH but some of the initial blood changes may be less severe and problems with clotting and the function of the heart may be worse.

Like other forms of HLH, viruses have been shown to trigger MAS, but also some medications. Those suffering from MAS have a better outcome than HLH, with a survival rate of 80-90%. Treatment is similar to HLH, but less intensive.

Symptoms

Children usually have a prolonged fever which may subside, but recurs after days or weeks. They have an enlarged spleen and liver as well as a reduced number of red and white blood cells and platelets. Some children may have fits.

If an infection triggered the HLH, then they may also have symptoms from this.

Diagnosis and Treatment

Diagnosis

In order to diagnose HLH, a number of tests may be carried out. These will include blood tests, a bone marrow examination, lumbar puncture, an MRI scan, chest x-ray and tests to look for viruses.

Children will also be screened for underlying immune deficiency syndromes and auto-immune diseases. If familial HLH is suspected, they will be investigated for one of the genes known to lead to HLH.

Treatment

The aim of the treatment is first to reduce the severe excessive inflammation that is responsible for the life-threatening symptoms.