

primary HLH affects **THE WHOLE FAMILY**

Your doctor has been trying to determine the cause of the illness affecting you or your loved one and thinks it may be primary HLH, also known as *familial HLH*. This diagnosis may feel scary or overwhelming, and it raises the question of whether other members of your immediate family might have primary HLH, too. One of the most important things you can do right now is to become aware of signs and symptoms that may be connected to primary HLH so you can look out for them.

HLH=hemophagocytic lymphohistiocytosis.



your doctor is testing for primary HLH

“Primary HLH” stands for primary hemophagocytic lymphohistiocytosis.

Keep in mind the possible signs and symptoms of primary HLH are not limited to those shown here, and not all patients may experience the same symptoms. Because of this—and because many of the signs and symptoms are common to other illnesses—primary HLH can be difficult to diagnose and is sometimes mistaken for other conditions, such as viral infections, sepsis, or cancer.

WHAT IS PRIMARY HLH?

Primary HLH is a rare genetic disease that generally affects infants and children, although it can affect adults as well. In primary HLH, the immune system, which normally defends the body against foreign invaders, does not work properly and attacks the person’s own cells and organs.

WHAT ARE THE SIGNS AND SYMPTOMS OF PRIMARY HLH?

Here is a list of possible signs and symptoms of primary HLH. Let your doctor know if you or your loved one have any of these or if any have worsened.

- High fever that lasts a long time
- Rash
- Seizures and neck stiffness
- Changes in mental state, such as confusion or memory loss
- Swollen or bloated stomach
- Swollen lymph nodes
- Very pale appearance
- Coughing and difficulty breathing
- Muscle weakness and trouble walking
- Problems with vision, such as blurriness
- Yellowing of the eyes and skin
- Stomachache, vomiting, or diarrhea
- Failure to grow or to gain or maintain weight (in infants and young children)

HOW DO PEOPLE GET PRIMARY HLH?

Primary HLH is caused by changes, or mutations, in a gene that affects how the immune system functions. The disease can be inherited, meaning that it can be passed down from parents to children, although not all genetic mutations that may cause primary HLH are known.

HOW CAN MY DOCTOR BE SURE IT’S PRIMARY HLH?

After checking for signs and symptoms like the ones listed on the left page and looking at the results of various blood tests, your doctor may be reasonably certain that you or your loved one have primary HLH. They can, however, use genetic tests to confirm. These tests may include:



Targeted individual gene sequencing

This is a medical tool that can help analyze specific mutations in a cell or tissue sample.

HLH gene panel testing

Gene panel testing is a laboratory test that studies the genes in a tissue sample.

Clinical WES

Most people who have WES (whole-exome sequencing) have already had some other genetic testing. WES is one of the most complete genetic tests available, and it may provide results that other genetic tests have not.

It’s important to know that even if a genetic test comes back negative, it is still possible that you or your loved one have primary HLH. Talk with your doctor about this when you get the results.

information for you and your family

Genetic counselors can help you understand the results of genetic testing. They specialize in providing information and support to families who are at risk for inherited conditions.

SHOULD THE REST OF MY FAMILY HAVE GENETIC TESTING?

Since primary HLH can be passed down from parents to children, it may be a good idea for other members of your immediate family to be tested. This is especially true if you or others in your family are pregnant or considering having children. Talk with your doctor or meet with a genetic counselor to discuss next steps for your family.

HOW LONG WILL IT BE UNTIL TREATMENT CAN BEGIN?

It can take several weeks to get the results of a genetic test. Your doctor may start treatment with conventional therapy before those results are available in order to manage the signs and symptoms that you or your loved one are experiencing. The goal is to get symptoms under control in preparation for a bone marrow transplant, the only curative treatment for primary HLH.

WHERE CAN I LEARN ABOUT BONE MARROW TRANSPLANT?

Your doctor is your best source of information. You may also find the following websites helpful:

bmtinfonet.org



bethematch.org

You are not alone in your primary HLH journey

There are a number of organizations that offer information, support groups, and a range of other services. Some of them were even started by families who may have experiences similar to yours.

Social workers specialize in helping families like yours

Most hospitals have social workers on staff to help families cope with diseases such as primary HLH. Ask your doctor, nurse, or hospital administrator to help you connect with one of them.

Note: All organizations listed here are third parties over whom Sobi has no control and are provided solely as resources. Sobi has no responsibility for the content provided by these third parties.

[Matthew and Andrew Akin Foundation \(matthewandandrew.org\)](http://matthewandandrew.org)

The mission of this foundation is to inform, inspire, and invest in families affected by HLH.

[Liam's Lighthouse Foundation \(liamslighthousefoundation.org\)](http://liamslighthousefoundation.org)

This organization works to increase awareness of HLH and other histiocytic disorders. It focuses on bringing affected families together, offering support through a variety of programs, and raising funds for research and education.

[HLH Support \(hlhsupport.org\)](http://hlhsupport.org)

Founded by the mother of an HLH survivor, the goal of this program is to support and connect with families struggling with the disease. In addition to an active Facebook support group, they hold events to raise money for HLH.

[Histiocytosis Association \(histio.org\)](http://histio.org)

This group provides educational materials and a physician directory for treatment or second opinions. They are also involved in advocacy and programs to raise funds for research.

[Immune Deficiency Foundation \(primaryimmune.org\)](http://primaryimmune.org)

Dedicated to improving the diagnosis, treatment, and quality of life for people with primary immunodeficiency diseases, this foundation provides advocacy, education, and research.

don't fight primary HLH alone

The primary HLH journey can be challenging. Take advantage of the support and resources available to you, and don't be afraid to ask questions.

