

## primary HLH affects

### **THE WHOLE FAMILY**

Finding out you or your loved one have primary HLH, also called familial HLH, can be scary. You may be wondering whether other members of your family could have it, too. This guide can help you learn which signs and symptoms to look out for. It also explains why some family members should be tested, even if they don't have any symptoms right now.

HLH=hemophagocytic lymphohistiocytosis.



# understanding primary HLH signs and symptoms

“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. Not all patients will have all these symptoms or the same symptoms. Because of this—and because many signs and symptoms are common to other illnesses—primary HLH can be difficult to diagnose. Sometimes, it may even be mistaken for other conditions, such as viral infections, sepsis, or cancer.

## WHAT IS PRIMARY HLH?

Primary HLH is a rare genetic disease that mostly affects infants and children, although it can affect adults, too. In primary HLH, the immune system, which normally defends the body against foreign invaders like viruses and bacteria, doesn't work properly. Instead, it 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 gotten worse.

- High fever that lasts a long time
- Rash
- Seizures or neck stiffness
- Changes in mental state, such as confusion or memory loss
- Swollen or bloated stomach
- Swollen lymph nodes
- Very pale appearance
- Coughing and trouble breathing
- Muscle weakness and trouble walking
- Problems with vision, such as blurriness
- Yellowing of the eyes and skin
- Stomachache, vomiting, or diarrhea
- Infant/child does not grow or gain weight

## HOW DO PEOPLE GET PRIMARY HLH?

Primary HLH is caused by changes, or mutations, in a gene that affects how the immune system works. The disease can be inherited, meaning that it can be passed down from parents to children.

## HOW CAN MY DOCTOR BE SURE IT'S PRIMARY HLH?

After checking for signs and symptoms and looking at the results of different blood tests, your doctor may say that you or your loved one have primary HLH. They can also use genetic tests to confirm. These tests check for mutations, or changes to DNA, known to cause primary HLH.

It is possible that you or your loved one have primary HLH even if genetic tests are negative. Talk with your doctor about this when you get the results.

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

## **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 family members to be tested. This is especially true if you have or are thinking about 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 before those results are available, since it is important to control symptoms quickly.

Most patients who have primary HLH will need a stem cell transplant to be cured. Getting symptoms under control will be important if the doctor decides transplant is best for you or your loved one.

## **You are not alone in your primary HLH journey**

There are many organizations that offer information, support groups, and other services. Some of them were even started by families like yours.

## **Social workers focus on 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 staff to help you connect with one of them.

## **[HLH Heroes Foundation \(ne-np.facebook.com\)](https://ne-np.facebook.com)**

Provides support to individuals and families impacted by HLH. This organization was created by family members and caregivers of people with HLH to create a community of support.

## **[HLH Support \(hlhsupport.org\)](https://hlhsupport.org)**

Created by the mother of a primary HLH survivor to help support and connect others. Their Facebook group provides a place for patients and families to share stories and ask questions. HLH Support also spreads awareness about resources and fundraisers.

## **[Histiocytosis Association \(histio.org\)](https://histio.org)**

Provides helpful information to patients and families dealing with primary HLH. It offers podcasts, webinars, videos, and more to help explain the condition. It can also help connect patients with doctors who have experience in treating primary HLH.

## **[Immune Deficiency Foundation \(primaryimmune.org\)](https://primaryimmune.org)**

Works to inform and connect families facing conditions like primary HLH. It hosts virtual meetings for people to share their experiences in a casual setting. The website offers lots of resources—including educational materials and fundraising information.

## **[Blood & Marrow Transplant Information Network \(bmtinfonet.org\)](https://bmtinfonet.org)**

Provides patients and their loved ones with emotional support and easy-to-understand information about stem cell transplants. Its website includes a video learning library, patient books, and more.

Note: Sobi is not responsible for the content provided by these third-party organizations.



# don't face 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.

