understanding primary HLH diagnosis and treatment

Diagnosing primary HLH can be difficult. Different doctors usually need to rule out other illnesses before they suspect this rare condition. If you or your loved one have been diagnosed, you may be wondering how the doctor figured it out. You may also have questions about starting treatment.

THIS GUIDE CAN HELP EXPLAIN:

1 How you or your loved one were diagnosed

2 The types of treatment the doctor may prescribe

HLH=hemophagocytic lymphohistiocytosis.

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how doctors diagnose primary HLH

Even if genetic testing comes back negative, it's still possible that you or your loved one may have primary HLH.

Other medical tests
Sometimes doctors run other tests to make sure that someone has primary HLH. Don't hesitate to ask whether any other tests were done.

GENETIC TESTING

Primary HLH is a genetic disease, which means that it can run in families. It is caused by changes, or mutations, in genes that affect how the immune system works. For this reason, doctors often use genetic tests to confirm whether someone has it.

Unfortunately, it may take several weeks to get genetic testing results. Since it's very important to treat primary HLH quickly, doctors may check for it another way.

DIAGNOSTIC CRITERIA

If a doctor thinks someone may have primary HLH, they can use a list of signs and symptoms to find out. This list is called "diagnostic criteria." A person who has **at least 5 of the 8** criteria may have primary HLH:

- Long-lasting fever
- Enlarged spleen
- Lower numbers of blood cells (cytopenias)
- A higher level of triglycerides (a certain type of fat cell) in the bloodstream and/or problems with clotting due to low levels of a protein called fibrinogen
- Higher levels of ferritin (a protein that stores iron)
- Evidence that blood cells might be damaging other blood cells (hemophagocytosis) in certain areas of the body
- Low levels of immune cells called "natural killer cells," or none at all
- Higher levels of sCD25, a molecule that increases with inflammation

To diagnose primary HLH, doctors must also rule out other illnesses like infections and cancers. These illnesses may have similar symptoms to primary HLH but should be treated differently.

how doctors start treating primary HLH

THERE ARE 2 MAIN TREATMENTS THAT DOCTORS USE TO BEGIN TREATING PRIMARY HLH

1 **A steroid called dexamethasone**

A drug that helps relieve swelling and inflammation

2 **A chemotherapy called etoposide**

A treatment, often used for cancer, that stops cell growth

Optional **An immunosuppressant called cyclosporine**

Sometimes, doctors may use cyclosporine in addition to dexamethasone and chemotherapy. It is a treatment that makes it easier for the body to accept a transplant

- Prevents the immune system from responding to new cells or tissues in a harmful way

Primary HLH is different for everyone. Keep in mind that the doctor may prescribe some or all of these treatments. Depending on how they work, he or she may change the treatment plan or consider other options.

You should know that these treatments were not specifically designed for primary HLH. They are not approved by the US Food and Drug Administration (FDA) to treat it. Doctors use them because they helped people who had primary HLH in clinical studies. However, they do not work for everyone.

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goals of primary HLH treatment

CONTROL SYMPTOMS

The symptoms of primary HLH are usually very serious. Your doctor will begin treatment as soon as possible to relieve them.

PREPARE FOR TRANSPLANT

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 a transplant is best for you or your loved one.

Keep in mind that it may take weeks or even months to calm the symptoms of primary HLH.

discussing diagnosis and treatment with your doctor

Once primary HLH has been diagnosed, there is a lot to take in. If you feel overwhelmed, asking questions is a great place to start.

HERE ARE SOME THINGS TO CONSIDER ASKING YOUR DOCTOR:

1. How did you diagnose primary HLH in me or my loved one?

2. Were there specific symptoms that made you think it was primary HLH?

3. Did you do genetic testing? What were the results?

4. Which types of treatments do you plan to use to treat primary HLH and why?

5. How long will each treatment be used?

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HERE ARE SOME THINGS TO CONSIDER ASKING YOUR DOCTOR:

6. How will you decide whether a specific treatment is working?

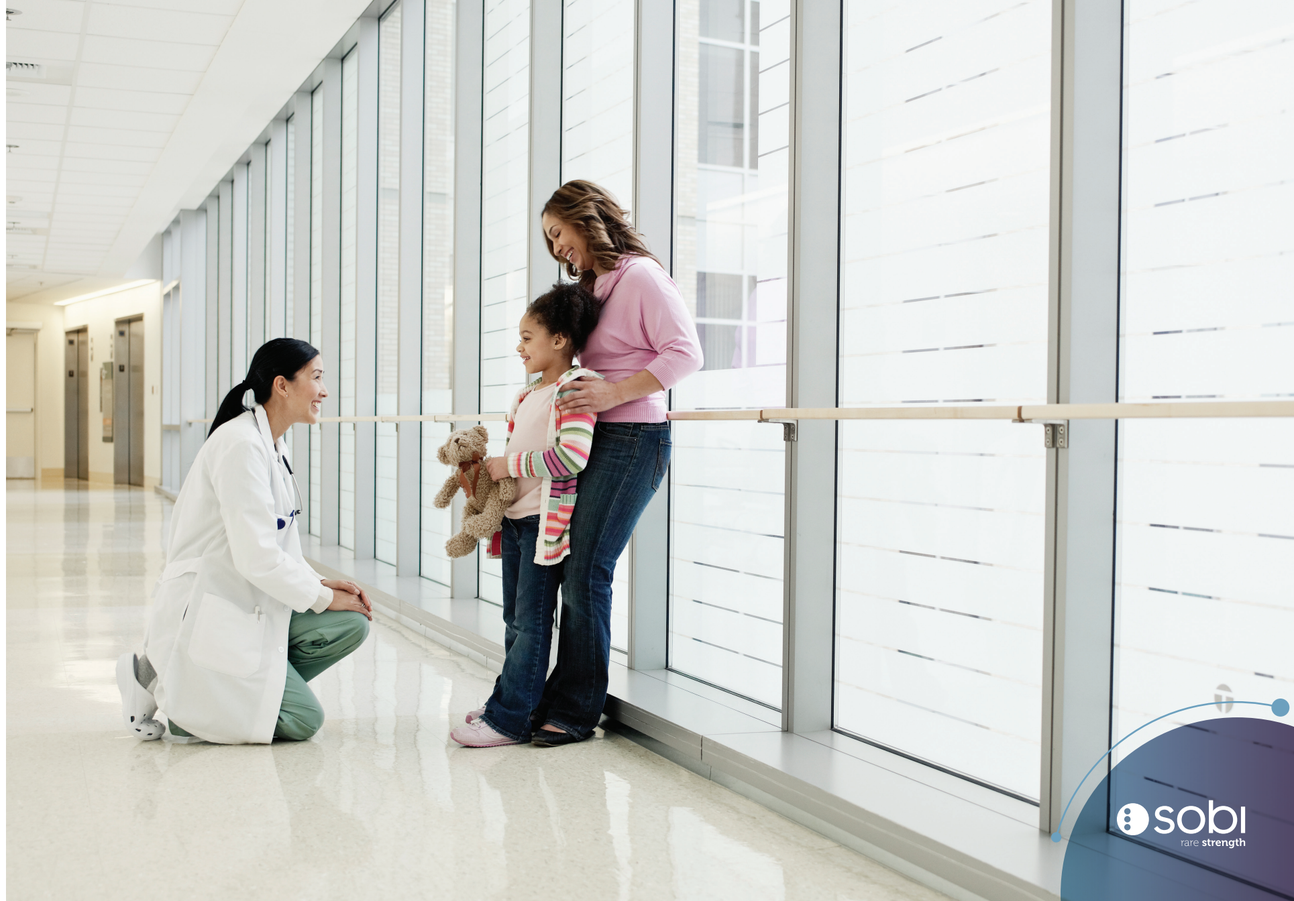
7. What are the side effects of each treatment you will prescribe?

8. What is the goal of treatment?

9. Do you think a stem cell transplant will be needed?

10. How long does treatment usually last?

11. Do you have any more information about stem cell transplant that you can share?



STARTING TREATMENT IS AN IMPORTANT STEP IN THE PRIMARY HLH JOURNEY.

We hope this guide helps you understand how primary HLH was diagnosed and how it may be treated.

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