

# *Texas Children's Cancer Center and Hematology Service*

## Hereditary Spherocytosis

This sheet will help you:

- learn about hereditary spherocytosis
- learn how hereditary spherocytosis is treated
- learn about things you can do to keep your child healthy if he or she has hereditary spherocytosis

### **What is hereditary spherocytosis?**

Hereditary spherocytosis (HS) is an inherited disease that causes anemia. If your child has hereditary spherocytosis, either parent may also have the disease. Occasionally, neither parent of an affected child has the disease; this is considered a spontaneous mutation. In the United States, approximately 1 in 5,000 people have hereditary spherocytosis.

Hereditary spherocytosis is a disease that results in the formation of abnormal red blood cells with fragile cell walls. Red blood cells circulate in the blood and contain hemoglobin, which carries oxygen to all parts of the body. Normal red blood cells are shaped like a disc. Spherocytes are round and fragile and do not change shape to pass through certain organs as easily as normal red blood cells. Because spherocytes cannot change their shape easily, they stay in the spleen longer than normal red blood cells, and the membrane surrounding the cell becomes

damaged. After circulating through the spleen many times, the cell eventually becomes so damaged that it is destroyed by the spleen.

### **What are the signs and symptoms of hereditary spherocytosis?**

Symptoms of hereditary spherocytosis vary depending on the severity of the disease. Many people with hereditary spherocytosis have a normal hemoglobin level. Most patients have only a mild anemia. These patients compensate by making more red blood cells, which is measured by the reticulocyte (immature red blood cell) count. However, infection, fever and stress can stimulate the spleen to destroy more red blood cells than usual. If this occurs, your child's hemoglobin level will drop and the bilirubin level may increase, causing the skin and whites of the eyes to be yellow.

Blood cells are produced in the bone marrow. Sometimes an infection (specifically, parvovirus) can cause the bone marrow to stop cell production temporarily. If this occurs, your child's hemoglobin will decrease, and there will be very few reticulocytes in the blood. This is called an aplastic crisis. It may take several days before red blood cell production will resume. Occasionally, anemia may be severe enough to require a red blood cell transfusion.

## What is the “osmotic fragility” test?

The osmotic fragility test is done to confirm the diagnosis of hereditary spherocytosis. A patient’s red blood cells are placed in different concentrations of saline solution for 24 hours. When red blood cells are placed in saline solution, they absorb water until the cell membrane bursts. Spherocytes do not tolerate weak saline solutions, causing them to burst sooner than normal cells.

## Treating hereditary spherocytosis

Splenectomy (surgical removal of the spleen) is the treatment of choice for symptomatic hereditary spherocytosis and can be done when a child is five years of age or older. Treatment before the age of five consists of daily folic acid (vitamin) supplementation.

Removing the spleen does not cure the disease, but it does allow the red blood cells to live longer so that a child no longer becomes anemic during periods of stress or infection and the skin and eyes do not turn yellow. After the spleen is removed, the child is at an increased risk for certain types of infection. For this reason, penicillin is given twice a day for the rest of your child’s life. It is very important that your child receive all of the normal childhood immunizations and a few special immunizations (pneumococcal and meningococcal immunizations) to prevent infection.

## When to call your health care provider

*Before splenectomy, call your health care provider or clinic right away if:*

- you notice that your child’s skin is more pale or yellow than usual and if the whites of the eyes are more yellow than usual
- your child has a fever of  $\geq 101.5^{\circ}$
- you are concerned about your child’s health.

*After splenectomy, call your health care provider or clinic right away if:*

- your child has a fever  $\geq 101.5^{\circ}$
- you are having trouble giving your child penicillin twice a day
- your child is pale or the skin and whites of the eyes are yellow
- you are concerned about your child’s health.

**\*Always tell your child’s health care provider that your child has hereditary spherocytosis. Also tell the health care provider whether or not your child has had a splenectomy.**

## Important phone numbers

Child’s doctor: \_\_\_\_\_

Child’s nurse practitioner: \_\_\_\_\_

Hospital: \_\_\_\_\_

Clinic: \_\_\_\_\_

Pharmacy: \_\_\_\_\_

24-hour pharmacy: \_\_\_\_\_

Other important numbers: \_\_\_\_\_

\_\_\_\_\_

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