Qure
Center for Complete Care
Blood Diseases, Cancer, Stem Cell Transplantation

Hereditary spherocytosis (HS)

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. In the United States, approximately 1 in 5,000 people have hereditary spherocytosis. The precise incidence in India is unknown.

HS results in the formation of abnormally fragile red blood cells. Red blood cells circulate in the blood and contain hemoglobin, which carries oxygen to all parts of the body, and normal red blood cells are shaped like a disc. However spherocytes are round and fragile and do not change shape to pass through certain organs (e.g. the spleen) as easily as normal red blood cells. Because spherocytes stay in the spleen longer than normal red blood cells, the membrane surrounding the cell becomes damaged, and eventually the red cell is destroyed there.

What are the signs and symptoms of HS?

Symptoms of HS vary, and many patients with HS have a normal hemoglobin level, or only mild anemia. Patients with HS 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, causing the hemoglobin level to drop and with jaundice of the skin and whites of the eyes.

Since red blood cells are produced in the bone marrow, sometimes a viral infection (e.g. parvovirus) can cause the bone marrow to stop cell production temporarily causing a drop in hemoglobin with lack of reticulocytes in the blood. This is called an “aplastic crisis” and may last for several days before red blood cell production will resume, during which anemia may be severe enough to require a red blood cell transfusion.

How is HS diagnosed?

The history and physical findings, and the appearance of “spherocytes “in the peripheral blood smear strongly the diagnosis. Confirmatory tests are:

- Osmotic fragility test: in which 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.
- EMA test: in which a patient’s red cells are stained with a special dye (EMA) looking for the protein (band 3) which is decreased or absent in spherocytes.

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How is HS treated?

Splenectomy (surgical removal of the spleen) is the treatment of choice for HS patients with significant symptoms, 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 underlying 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 decrease the risk of infection.

When should I call my doctor?

Call the clinic right away if:

- you notice that your child's skin is more pale than usual or he or she seems more tired.
- your child's skin and whites of the eyes are more yellow than usual
- your child has a fever of >100.5° F