

Hereditary Spherocytosis (HS)

What is hereditary spherocytosis (HS)?

Hereditary spherocytosis (HS) is a medical term for a condition which affects the walls of the red blood cells. They get stuck in an organ called the spleen, which then destroys them. The process where red blood cells are destroyed is called *hemolysis*.

What are red blood cells?

Blood contains 3 types of cells: red blood cells (RBC), white blood cells (WBC), and platelets. Red blood cells are the most common type of blood cells, and are responsible for delivering oxygen to all parts of the body.

Red blood cells, like white cells and platelets, are continuously made in the bone marrow. When released into the bloodstream, the average lifespan of a red cell is 120 days.

Under the microscope, normal red cells are shaped like discs or donuts with the center partially scooped out. This shape makes them very soft and flexible, so they can easily squeeze through very small blood vessels.

Sometimes there is a problem with the wall of the red cell which changes shape to look like sphere or ball. These cells are called *spherocytes*. They are not as soft or flexible as regular red blood cells. As a result, they have difficulty moving through small blood vessels, and tend to get stuck.

How is HS diagnosed?

HS is diagnosed by medical evaluation and blood tests. These blood tests include a *complete blood count* (CBC). If the medical evaluation and CBC are suspicious for HS, special blood tests and scans are needed.

How did my child get HS?

HS is usually inherited from one parent, but this is not always the case. Sometimes people with HS may not know they have the illness unless they undergo testing.

HS is found in about 1/5,000 people. It is more common in people of Northern European ancestry, but is found in other ethnic groups.

How severe are the symptoms of HS?

The symptoms of HS vary from person to person, and can be mild, moderate or severe. Mild HS (about 20% of cases) will have minimal symptoms and may not be diagnosed until adulthood. Severe HS (about 5% of cases) is usually diagnosed in newborns and infants, and requires very careful medical attention. Moderate HS (about 75% of cases) is somewhere in between.

What are the symptoms of HS?

Symptoms of HS are due to 2 processes: hemolysis and anemia.

When red blood cells break down (*hemolysis*), they release bilirubin into the bloodstream, which causes yellowing of the skin (*jaundice*) and eyes. Hemolysis causes different problems depending on the age:

- Newborns – may need light therapy or further measures
- Children – increasing size of the spleen
- Teens/adults – gallstones that may require surgery

If enough red blood cells are destroyed, the red blood cell count will be low (*anemia*). Symptoms of anemia include looking pale, being tired or weak, headaches, poor concentration, and challenges with behavior and school.

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How is HS treated?

There is no cure for HS. Folic acid supplement should be taken. Folic acid is an important building block for red blood cells and is needed in higher amount when red blood cells are being destroyed.

Other treatments may include:

- Blood transfusions
- Removal of part or all of the spleen

What should I watch for?

Certain viral infections can cause the bone marrow to be paralyzed, or cause the spleen to destroy more red blood cells than usual. This can cause a dangerously low level of red blood cells.

Increasing paleness or yellowness, especially if accompanied by low energy, difficulty breathing, headaches or irritability are some of the signs that the red cell count is dangerously low.

If your child's spleen is enlarged, contact sports and other high risk activities must be avoided. This is because the spleen can rupture if it is hit. Sometimes a spleen guard can be worn for protection.

For more information you may contact the Hematology Department at BC Children's Hospital:

Hematology Clinic: (604) 875-2116

Hematology Nurse Clinician
Susan Lee, RN
(604) 875-2345 ext 7060
Email: slee@cw.bc.ca

After hours/weekend/emergency
Hematologist on-call: (604) 875-2161