The Blood Project

UNDERSTANDING HEREDITARY SPHEROCYTOSIS

A brief guide for patients with HS

Hereditary spherocytosis (HS) is a genetic condition that affects red blood cells. In HS, red blood cells are shaped differently and break down more quickly than normal, leading to anemia and jaundice. Many people have a mild form and live full, active lives with monitoring and simple treatments, while others need closer follow-up.

What are red blood cells?

Red blood cells carry oxygen from the lungs to the rest of the body.
Normal red blood cells are shaped like flexible discs, which allows them to squeeze through small blood vessels and circulate for about 120 days before being removed.

What is hereditary spherocytosis?

In hereditary spherocytosis, red blood cells are rounder and less flexible than normal.
These cells are called *spherocytes*.
Because they cannot bend easily, they are trapped and removed early by the spleen.
This leads to chronic breakdown of red blood cells, called *hemolysis*.

Why it happens

Hereditary spherocytosis is caused by inherited changes in proteins that support the red blood cell membrane, such as spectrin, ankyrin, band 3, or protein 4.2.
These proteins normally help red blood cells keep their flexible disc shape.
The condition is most often inherited in an autosomal dominant pattern, meaning one affected parent can pass it on.
In some cases, HS occurs because of a new genetic change, even without a family history.
Because of this inheritance pattern, family members may be offered testing or evaluation, especially children or siblings.

Does it cause symptoms?

Symptoms vary widely and depend on how severe the red blood cell breakdown is.
Some people have no symptoms, while others develop anemia or jaundice.

Symptoms in newborns and infants (neonatal jaundice)

In newborns, HS often causes jaundice in the first days of life.
This jaundice may be more intense or last longer than typical newborn jaundice and requires close monitoring.
Some infants develop anemia in the first months of life and may need phototherapy or blood transfusions.
Early monitoring helps prevent complications.
Parents should contact a doctor urgently for:

- increasing yellowing of the skin or eyes
- poor feeding or extreme sleepiness

- pale skin or fast breathing

Symptoms in children and adults

Symptoms may include:

- fatigue or low energy
- pale skin
- yellowing of the eyes or skin
- darker urine
- abdominal discomfort from an enlarged spleen
- episodes of worsening anemia during illness

As children grow, the focus often shifts to monitoring anemia, spleen size, and gallstones.

Is it dangerous?

Most people with HS have mild to moderate disease and do well with routine care.
However, some complications require prompt attention.

Aplastic crisis

An aplastic crisis occurs when the bone marrow temporarily stops making red blood cells.
This is most often triggered by parvovirus B19 infection.
Because red cells continue to break down, hemoglobin can fall quickly.
This can be life-threatening and usually requires urgent blood transfusion.
Warning signs include sudden severe fatigue, marked paleness, rapid heartbeat, or shortness of breath.

Hemolytic crisis

A hemolytic crisis refers to a sudden increase in red blood cell breakdown, often during infection or stress.
This leads to worsening anemia and jaundice.
Symptoms may include darker urine, increased yellowing, fatigue, and abdominal discomfort.
This usually improves as the illness resolves.

Gallstones

Gallstones are common in HS because chronic red blood cell breakdown increases bilirubin levels.
Over time, bilirubin can form pigment gallstones.
Gallstones may cause right-sided abdominal pain, nausea, vomiting, or gallbladder infection.
Many people have gallstones without symptoms, but monitoring is important.

Problems from an enlarged spleen

An enlarged spleen may cause abdominal fullness or discomfort.
Very large spleens increase the risk of red blood cell destruction and, rarely, injury with contact sports.

How your doctor evaluates it

Evaluation usually includes:

- complete blood count (CBC)
- reticulocyte count
- bilirubin and other markers of hemolysis
- a blood smear showing spherocytes
- specialized tests (such as EMA binding or osmotic fragility)
- abdominal ultrasound to check for gallstones

Family history often supports the diagnosis.
Genetic testing may be considered in selected cases.

What Is the treatment?

Treatment depends on severity.

Mild HS

Often requires folic acid supplementation and monitoring.
Folic acid helps the bone marrow keep up with increased red blood cell production.

Moderate HS

May require closer follow-up and occasional blood transfusions during illness or stress.
Gallstones and spleen size are monitored over time.

Severe HS

May require more frequent transfusions and consideration of splenectomy if anemia or symptoms are difficult to manage.
Splenectomy can reduce anemia by preventing early red blood cell destruction.
However, it increases lifelong risk of serious infections.
This decision is made carefully with a hematologist and, for children, their family.
Vaccinations and long-term precautions are essential.
Partial splenectomy may be an option in selected patients to preserve some immune function.

Treating gallstones

If gallstones cause symptoms, gallbladder removal may be recommended.
Gallbladder surgery is sometimes performed at the same time as splenectomy.

When should I contact my doctor?

Contact your doctor for:

- worsening fatigue or pallor
- increasing jaundice or dark urine
- persistent or worsening abdominal pain
- signs of infection

Go to the emergency department immediately for:

- fever after splenectomy
- sudden severe weakness, shortness of breath, or rapid heartbeat

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What is the usual plan going forward?

Most people with hereditary spherocytosis are followed long term with periodic blood tests and clinic visits.
The focus is on monitoring anemia, preventing complications, managing gallstones, and reassessing treatment needs over time.
With appropriate care, most people live full, active lives.

Key points to remember

- **hereditary condition:** HS affects red blood cell shape and survival
- **often mild:** many people have stable disease with few symptoms
- **jaundice and anemia:** common features, especially in infancy
- **watch for crises:** aplastic and hemolytic crises need urgent care
- **splenectomy decisions:** helpful for some, but with lifelong infection risk
- **long-term outlook:** with monitoring and care, most people do very well