

# Hereditary Spherocytosis

Hereditary spherocytosis (HS) is an inherited condition of red blood cells. The disease can be mild and go unrecognised in some people. In others there may be severe anaemia requiring regular blood transfusions.

Some people with hereditary spherocytosis may be offered surgery to remove their spleen. This usually helps the condition, but increases their risk of developing severe infections.

## What is hereditary spherocytosis?

Hereditary spherocytosis (HS) is an inherited condition that affects your red blood cells. The red blood cells are those that carry oxygen around the body. Their shape (like a slightly elongated saucer) helps them carry oxygen effectively.

In HS there is a defect in the outer layer of the red blood cell. This means the cell is not the usual shape. Spherocytosis means more sphere-shaped and this also makes it more fragile than usual.

## How common is hereditary spherocytosis?

In the USA and Europe, HS is found in about 1 in 2,000 people. There are many different types of defect but they all produce similar problems.

## What are the features of hereditary spherocytosis?

The features of hereditary spherocytosis are usually:

- Anaemia.
- Yellowing of your skin and of the whites of your eyes (jaundice).
- A large spleen.

These features can happen at any time of life.

Anaemia means that you have fewer red blood cells than normal, or you have less haemoglobin than normal in each red blood cell. Anaemia can make you feel tired and short of breath with little or no exercise.

Jaundice can occur for many different reasons. For example, a common reason for jaundice is because the liver isn't working properly.

In hereditary spherocytosis you become jaundiced because your red cells are broken down too quickly. As they are broken down, the red cells release a coloured substance – a pigment. The body cannot clear it away quickly enough and it shows in your skin and eyes.

In hereditary spherocytosis the spleen has to work harder than usual. This means the spleen grows and becomes larger than normal.

## How severe is hereditary spherocytosis?

As regards severity:

About 2-3 in 10 people with HS have mild disease. They will be losing red blood cells but their body will be able to replace them quickly. They may have no symptoms.

About 6-7 in 10 people with HS have moderate or severe disease. Half of these are first diagnosed in childhood with anaemia.

## How is hereditary spherocytosis inherited?

Hereditary spherocytosis can be inherited in three different ways, depending on the exact type of HS present.

### Autosomal dominant

This means that you can inherit HS from just one of your parents. If one of your parents has a faulty copy of the gene, there is a 50:50 chance that each child they have will inherit the faulty gene and develop HS. This is the most common way to inherit HS. About 7 out of 10 cases are inherited this way.

### Autosomal recessive

This means that, in order to develop HS, you need to inherit two abnormal genes – one from your mother and one from your father. If you inherit only one abnormal gene, you are called a carrier. Carriers do not have the disorder, as they have one normal gene which is enough to make normal-shaped red blood cells. However, carriers can pass the abnormal gene on to their children.

### New mutation

Occasionally, however, someone with HS may not have a history of the condition in their family. This may be because of what is called a new mutation, or a de

novo mutation. A new mutation is a fault in a gene that is present for the first time in one family member. It can happen because of:

A fault in the genetic material in either the egg or sperm of one of the affected person's parents; or

A fault in the genetic material of the fertilised egg when the egg and sperm meet (the embryo).

It is not clear what causes this mutation suddenly to occur for the first time.

If you are diagnosed as having HS, it might be useful to see a genetic counsellor. They can advise you about the specific risks of passing on your type of HS if you have children.

## How is hereditary spherocytosis diagnosed?

The changes in the red blood cell can be seen through a microscope. They will be noticed if a blood sample is sent to the laboratory. Another cell (a reticulocyte) may also be seen. This shows that the red blood cells are being broken down too quickly.

Other blood tests are sometimes needed. They help to rule out other causes of red blood cell changes, such as liver disease, zinc toxicity and some infections.

Your doctor may also notice that your spleen is larger than usual, if they examine your tummy (abdomen).

## Hereditary spherocytosis treatment

Treatment for hereditary spherocytosis typically include:

No treatment (for mild cases).

Blood transfusions if there is severe anaemia.

Folate tablets (or liquid for small children) – these are also given to help the body to keep replacing the red blood cells. A normal diet will not meet the increased demand for folate.

Removing the spleen.

Medicines such as steroids may reduce the need for transfusions in a crisis – see below.

## Removing the spleen

Removing the spleen (splenectomy) is known to help control the disease. The blood cells are able to survive for much longer in the bloodstream if the spleen is removed. The decision to do this is not straightforward though. Removing the spleen means the risk of a life-threatening infection is increased for that person.

After a splenectomy, people with hereditary spherocytosis will require lifelong preventative antibiotics – such as low-dose penicillin or erythromycin. It may also be sensible for them to have a full treatment dose of antibiotics at home, in case an infection develops. This will mean treatment will start as soon as possible and not depend, for example, on pharmacy opening hours.

People with HS who have had their spleen removed will also require vaccinating against various infections.

Because of these risks, the person with HS will need to understand how to recognise that an infection is developing. Carrying a card or wearing a special medical emergency bracelet or similar that explains the person has had a splenectomy, is also sensible.

Research is showing that just removing part of the spleen may also be effective. More evidence is needed.

## Hereditary spherocytosis complications

### Gallstones

Gallstones are often found in people who have HS. The gallbladder is often removed at the same time as the spleen, if you also have symptoms of gallstones. Gallstones usually cause pain under the ribs on the right side after a fatty meal.

### Worsening of symptoms

A sudden worsening of symptoms can occur which doctors call a crisis. In HS a crisis can cause yellowing of your skin or the whites of your eyes (jaundice), anaemia and a tender spleen very quickly.

They can be triggered by viral infections such as colds and flu. Painkillers and plenty of fluids (possibly given through the vein) are usually all that is needed.

One particular infection – parvovirus B19 – may suppress the bone marrow. This can be life-threatening and usually lasts for 10–14 days. A crisis can also be caused by a lack of folate.

## **What is the outlook (prognosis) for hereditary spherocytosis?**

Most children have mild disease and do not need removal of the spleen (splenectomy). They have a normal life expectancy. If the spleen is removed, the red blood cells will survive for much longer and fewer transfusions are then needed. However, there is an increased risk of life-threatening infections.