Evelina London

NHS
Guy's and St Thomas'
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Hereditary spherocytosis

Information for patients and their families

showing we care

What is hereditary spherocytosis?

Hereditary spherocytosis (HS) is a condition affecting the red blood cells. Red blood cells help to carry oxygen around the body.

All the cells in our body have an outer layer called a membrane. Membranes protect the cell and allow substances to pass in and out. In HS the membrane of the red blood cells is fragile. This is because there is an abnormal protein on the surface of the red cell. When the red cells pass through the spleen, bits of membrane containing the abnormal protein are removed. This changes the shape of the red cell (which normally looks like a doughnut) to a sphere (spherocytic red cells).

Spherocytic red cells may only last 30-60 days, compared to normal red cells which last 120 days. Sometimes the bone marrow (the factory of the blood) cannot keep up with making enough red blood cells and the person may become anaemic (low red blood cells).

How common is HS?

Around one in 5,000 people has HS.

What causes HS?

HS is a genetic condition usually passed from parents to their children. It is more common in people of Northern European descent, but it is also common in North Africa, Japan, Brazil and can occur in any ethnic group.

Three out of four people affected by HS have a family history of HS. If a parent has HS, each of their children has a one in two chance of having the condition.

One in four people affected by HS don't have a family history of HS. This is because one of the parents may have a very mild condition and so have not been diagnosed, or the HS has been caused by a new genetic mutation.

What are the symptoms of HS?

People experience symptoms differently. HS is not always noted from birth and some people with very mild disease may not be aware they have it. Children often have similar symptoms to their affected parents or siblings.

There are four common problems associated with having HS.

1. **Anaemia** is a low red blood count. The symptoms of anaemia can be common in young children. Children may seem tired, irritable, not eat or feed well, have a pale colour and may not grow as well as expected. Older children and adults may get tired easily with exercise. How severe anaemia is will vary between different families. Some will have no symptoms, while others will need frequent red cell transfusions.

   Sometimes viral infections (and particularly an infection with the virus parvovirus B19, which causes slapped cheek syndrome) may stop production of red cells for up to 10 days. This can cause the anaemia to become a lot worse in a few days and patients may require a blood transfusion at that time. This is called an **aplastic crisis**.

2. **Jaundice** is caused by a substance called bilirubin which is released when red cells break down. People with HS usually have more bilirubin in their blood. This high level causes a yellow colour in the skin and the eyes.

3. **Gallstones** are small stones that form in the gallbladder, a small sack under the liver. The gallstones are caused by substances that build up when the red blood cells break down. They can occur in children as young as 10. They can cause abdominal (tummy) pain, inflammation of the gallbladder, blockage of bile and worsening jaundice. The main treatment for symptomatic gallstones is removal of the gallbladder, which requires surgery. Sometimes another procedure can widen the outlet of the gallbladder.

4. **Splenomegaly (large spleen**). The spleen is an organ that filters the blood, destroys old red blood cells and helps the body fight infections. People with HS may have large spleens, because the spleen is busy removing the fragile red cells. Some people who have large spleens or severe anaemia may need to have their spleens removed with an operation called a splenectomy.

How is HS diagnosed?

HS is diagnosed with a blood test. It is also important for the doctor to examine patients with HS and talk to the patients and their families.

Is HS contagious?

No, HS is not contagious.

How is HS treated?

Most children with HS do not need to do much about their condition at all. The bone marrow has the ability to increase the number of red blood cells it produces and so is able to keep up replacing the fragile red cells that have been removed.

A few things may be helpful:

1. **Folic acid**
   We encourage all patients to take folic acid as it helps the bone marrow replace the fragile blood cells.

2. **Clinic reviews**
   People with HS visit clinic at least once a year. They will occasionally require blood tests to check for the level of anaemia and red cell breakdown (haemolysis).

3. **Abdominal ultrasounds**
   An abdominal ultrasound might be needed if there is frequent abdominal (tummy) pain to check for the development of gallstones.

4. **Splenectomy**
   Removal of the spleen (splenectomy) stops red blood cells from breaking down and solves the problems of HS.

The spleen is an important organ of the immune system and patients who do not have a spleen may be prone to getting serious infections. This is higher in younger children, but may be less than previously expected because of newer vaccinations. Splenectomy is avoided in children younger than six years of age.

After removal of the spleen, people are at increased risk of infections. To decrease the risk of these infections people with HS who have had a splenectomy receive extra vaccinations and are placed on prophylactic antibiotics lifelong. They are also advised to seek medical attention urgently when they feel unwell or have high temperatures.

Will my child require a splenectomy?

Removal of the spleen (splenectomy) was used a lot in the past for patients with HS. It stops red blood cells from breaking down and solves the problems of HS. It is only advised for people who are experiencing a lot of symptoms from the HS (significant tiredness which interferes with daily activities, abdominal pain from an enlarged spleen, poor growth, or severe anaemia that requires frequent red blood cell transfusions).

Some surgeons may offer a partial splenectomy, where only part of the spleen is removed in the hope that fewer red cells will be destroyed and the patient will still have the protection from infections. This procedure has not been studied in a clinical trial and not many surgeons are experienced in this procedure.

What is the outlook for children with HS?

HS is a lifelong condition. Many people with this condition will experience few problems and some may not be aware that they have it. Some people may experience frequent and severe symptoms and may require red cell transfusions and surgical procedures such as gallbladder removal (cholecystectomy) and spleen removal (splenectomy). It is expected that with good medical care all patients should enjoy a full life.

When to seek help

You should bring your child to the hospital **immediately** in the following circumstances:

- sudden lethargy – they are hard to wake up or seem confused
- poor appetite/feeding
- jaundice or paleness that gets worse.

Contact details

Paediatric Haematologist: 020 7188 6203

Contact us

Evelina London Medicines Helpline
If you have any questions or concerns about your child's medicines, please speak to the staff caring for them or contact our helpline.
t: 020 7188 3003 10am to 5pm, Monday to Friday
e: letstalkmedicines@gstt.nhs.uk

Your comments and concerns
For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS). To make a complaint, contact the complaints department.
t: 020 7188 8801 (PALS) **e:** pals@gstt.nhs.uk
t: 020 7188 3514 (complaints) **e:** complaints2@gstt.nhs.uk

Language Support Services
If you need an interpreter or information about your care in a different language or format, please get in touch:
t: 020 7188 8815 **e:** languagesupport@gstt.nhs.uk

NHS Choices
Provides online information and guidance on all aspects of health and healthcare, to help you make choices about your health.
w: www.nhs.uk

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