

# Hereditary spherocytosis

## Information for patients

### What is hereditary spherocytosis?

Hereditary spherocytosis (HS) is an inherited condition affecting red blood cells. Red blood cells contain haemoglobin that transports oxygen around the body.

All the cells in our body have a surface membrane, which is a layer that controls the movement of substances in and out of the cell. In HS the surface membrane of red blood cells is less stable. When the red blood cells pass through the spleen, bits of the membrane are removed. This changes the shape of the red blood cells from doughnut-shaped discs to spheres. Doctors call these cells spherocytic red blood cells.

Spherocytic red blood cells are easily destroyed and may last only 30 days, compared to 120 days for normal red blood cells. Often the patient's bone marrow cannot keep up with making enough red blood cells and the person may become anaemic.

The breakdown products of the red blood cells can build up in the blood, causing an increase in the level of a chemical called bilirubin. Increased levels of bilirubin can make the whites of the eyes and skin turn yellow, a condition called jaundice. Over time the high levels of bilirubin can solidify in the bile (liquid produced by the liver, which helps to digest fats), forming gallstones.

### Why have I/has my child got hereditary spherocytosis?

HS is a genetic condition usually passed from parents to their children. It is more common in people of Northern European descent, but is also common in North Africa, Japan and 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 one in two chances of having the condition.

One out of four people affected by HS do not have a family history of HS. This is either because the parents have a very mild condition and so have not been diagnosed, or HS has been caused by a new genetic mutation.

### What are the symptoms of hereditary spherocytosis?

The severity of symptoms varies, but children often have similar symptoms to affected parents or siblings. Symptoms of HS can appear at any age and is not always noted from birth.

## **Anaemia**

Symptoms of anaemia can be quite vague in young children. The child may be lethargic, irritable, feed poorly, appear pale, or may not grow as well as expected. Older children and adults may tire when they exercise. The degree of anaemia varies between different families, from mild with no symptoms, to severe needing regular transfusions.

Sometimes viral infections (for example, slapped cheek syndrome or parvovirus B19) may cause the anaemia to become a lot worse very quickly. This may last for up to 10 days. If you develop sudden signs of anaemia, it is important that you seek medical attention for a blood test. Sometimes a short period of blood transfusion may be necessary.

## **Jaundice**

The symptoms of jaundice are the yellowing of the skin and the whites of the eyes. Jaundice is reversible and may vary over time.

## **Gallstones**

Gallstones are small stones that form in the gallbladder, a small pouch underneath the liver. They can cause recurrent pain in the abdomen, inflammation of the gallbladder or blockage of the bile, worsening jaundice. The main treatment is removal of the gallbladder, but gallstones can sometimes be managed with a widening of the outlet of the gallbladder.

## **Splenomegaly (large spleen)**

The spleen is an organ that acts mainly as a filter for the blood, destroying old blood cells and fighting infection. It is normally hidden under the ribs on the left hand side of the chest. If a spleen gets large, it will protrude into the abdomen and make it feel full or swollen. The spleen has a large blood supply and if damaged it can bleed very heavily.

If you have a large spleen, you should not play contact sports and if you are in an accident you should go to an emergency department (A&E) to be examined and scanned if necessary. Sometimes people who have large spleens, or have problems with anaemia, may need to have their spleens removed with an operation called a splenectomy.

## **How is hereditary spherocytosis diagnosed?**

To diagnose HS the doctor will ask some questions, perform an examination and do some blood tests.

## **What are the treatment options for HS?**

We will see you or your child in clinic at least once a year. We encourage all patients to take the vitamin folic acid as it helps the bone marrow to replace all the broken down red blood cells.

Some severely affected people may need their spleen removed (splenectomy). It is preferable to wait until a patient is six years old before doing this.

## When should I seek medical help?

If you notice any of the following symptoms, please go to your local emergency department (A&E):

- sudden lethargy
- poor appetite/feeding
- worsening jaundice or paleness.

A large spleen is more at risk of bleeding after being knocked. Seek urgent medical advice if you or your child receive an injury to the abdomen or are suffering from abdominal pain.

After removal of the spleen people are at increased risk of infections. They have extra vaccinations and take a preventative (prophylactic) dose of penicillin daily for life. For this reason, you should take infections, fevers or animal bites seriously and seek medical advice as you may need prompt treatment with antibiotics.

## Contacts

### Haematology consultants

Dr Bernard Davis  
Dr Farrukh Shah  
Dr Ali Rismani

### Paediatric consultants

Dr Andrew Robins  
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### Patient advice and liaison service (PALS)

If you have a question, compliment, comment or concern please contact our PALS team on 020 7288 5551 or [whh-tr.whitthealthPALS@nhs.net](mailto:whh-tr.whitthealthPALS@nhs.net)

If you need a large print, audio or translated copy of this leaflet please contact us on 020 7288 3081. We will try our best to meet your needs.

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Date published: 24/11/2015  
Review date: 24/11/2017  
Ref: MFN/Thal/HdSp/01

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