Patient Information Leaflet

York and Scarborough Teaching Hospitals NHS Foundation Trust

Hereditary Spherocytosis

Information for patients, relatives and carers

ⓘ For more information, please contact:

Ward 18, York Hospital, telephone 01904 726 018

Rainbow Ward, Scarborough Hospital, telephone 01723 342 336

Or email yhs-tr.childhealthadminyork@nhs.net

What is hereditary spherocytosis?

Hereditary spherocytosis (HS) is an inherited condition (passed down from parents) 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 or spherocytes. 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. 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 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 brothers and sisters. Symptoms of HS can appear at any age and are not always noted from birth.

Anaemia

Symptoms of anaemia can be quite vague in young children. The child may be more tired than usual, 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.

Parvovirus B19 (Slapped Cheek)

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

Jaundice

The symptoms of jaundice are 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 and 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, its lower border will push 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. The diagnosis can be difficult in the first few months of life, so blood tests may need to be repeated when the child is six months or older to confirm the diagnosis.

What are the treatment options for hereditary spherocytosis?

We will see you or your child in clinic at least once a year. Some patients with rapid cell breakdown are encouraged to take the vitamin folic acid as it helps the bone marrow to replace 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.

Plan for infants born to a parent with hereditary spherocytosis

If the mother or father have HS there is a one in two chance their child will be affected. Even if the parent has mild HS there is a chance that the baby may develop severe anaemia and/or jaundice in the first few weeks of life. It is important the midwife and/or obstetrician know about the HS so that a plan can be made following birth.
This should include blood tests for the baby within 12 hours of life. It is often difficult to confirm HS in the first few months of life so the tests may need repeating to monitor the haemoglobin and jaundice level.

When should I seek medical help?

If you notice any of the following symptoms, please go to your local emergency department (A&E) or call Ward 18 / Rainbow Ward:

- Sudden extreme tiredness
- 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 may take a preventative (prophylactic) dose of penicillin twice daily for life.
For this reason, you should take infections, fevers or animal bites seriously and seek medical advice as you or your child may need prompt treatment with antibiotics.

Vaccinations

Children with HS can and should receive all nationally recommended vaccinations.

Tell us what you think of this leaflet

We hope that you found this leaflet helpful. If you would like to tell us what you think, please contact: Dr Rebecca Proudfoot, Consultant Paediatrician, York Hospital, Wigginton Road, York, telephone main switchboard 01904 631313 or email yhs-tr.childhealthadminyork@nhs.net.

Patient Advice and Liaison Service (PALS)

PALS offers impartial advice and assistance to patients, their relatives, friends and carers. We can listen to feedback (positive or negative), answer questions and help resolve any concerns about Trust services.

PALS can be contacted on 01904 726262, or email yhs-tr.patientexperienceteam@nhs.net.

An answer phone is available out of hours.

Teaching, training and research

Our Trust is committed to teaching, training and research to support the development of health and healthcare in our community. Healthcare students may observe consultations for this purpose. You can opt out if you do not want students to observe. We may also ask you if you would like to be involved in our research.

Leaflets in alternative languages or formats

If you would like this information in a different format, including braille or easy read, or translated into a different language, please speak to a member of staff in the ward or department providing your care.

Patient Information Leaflets can be accessed via the Trust's Patient Information Leaflet website: www.yorkhospitals.nhs.uk/your-visit/patient-information-leaflets/

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