Hereditary spherocytosis

North Central London Haemoglobinopathy Network jointly with Whittington Health, Royal Free London, and Luton and Dunstable NHS Foundation Trust

Joint Red Cell Unit
If you need a large print, audio, braille or translated copy of this booklet, please contact us on 020 3447 9638. We will do our best to meet your needs.

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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 – 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 donut-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 chance of having the condition.

One out of four people affected by HS don’t 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. We are now able to analyse the DNA of the family members to understand what gene is causing the condition and find out who is affected.

What are the symptoms of hereditary spherocytosis?

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

**Anaemia**

The symptoms of anaemia can be very non-specific 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 parovirus B19, also known as slapped cheek syndrome) may stop the production of red cells for up to 10 days. This can cause the anaemia to become a lot worse over a few days. If you develop 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. For more information, please see our leaflet ‘Parovirus and haemolytic anaemias’.
Jaundice
The symptoms of jaundice are the yellowing of the skin and the whites of the eyes. Jaundice is reversible and may vary in intensity over time.

Gallstones
Gallstones are small stones that form in the gallbladder, a small pouch underneath the liver. They can cause recurrent pain 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 folic acid as it helps the bone marrow to replace all the broken down red blood cells.
- Some patients who are severely affected by HS may need to have their spleen removed (splenectomy). It is generally recommended to wait until a patient is five years old before doing this.

Some surgeons offer a partial or near-total splenectomy in the hope that fewer red cells will be destroyed and you will still have some spleen function left. This is important as the spleen can protect you against certain infections. However, this has never been confirmed in a clinical trial and so the results are still inconclusive.

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 and so have extra vaccinations and take a preventative (prophylactic) dose of penicillin lifelong. For this reason, you should take infections, fevers or animal bites seriously and seek medical advice as you may need prompt treatment with antibiotics.

If you are coming to A&E for any of these reasons, let us know using the numbers on the next page. This is not essential but will help us to streamline your care.
Contact details

Haematology advice line (office hours, adults and children):
020 3447 7359

Adult haematology advice line (out of hours):
07852 220 900

Paediatric helpline (out of hours):
  • nurse in charge 07961 081 645
  • ward T11 south 020 3456 7890 ext. 71103 or 71143

Apheresis:
020 3447 1803

Address: The Joint Red Cell Unit
  Department of Haematology
  3rd Floor West, 250 Euston Rd
  London NW1 2PG

Website: www.uclh.nhs.uk/JRCU

Haematology consultants:
Professor John Porter
Dr Sara Trompeter
Dr Perla Eleftheriou
Dr Emma Drasar
Dr Bernard Davis
Dr Farrukh Shah

Specialist nurses:
Bernadette Hylton (adults)
Nancy Huntley (apheresis)
Nina Gorman (children)
Where can I get more information?

Sickle Cell Society
Tel: 020 8861 7795
Website: www.sicklecellsociety.org

NHS Sickle Cell and Thalassaemia Screening Programme
Website: www.gov.uk/guidance/sickle-cell-and-thalassaemia-screening-programme-overview

UK Thalassaemia Society
Tel: 020 8882 0011
Fax: 020 8882 8618
Email: office@ukts.org
Website: www.ukts.org

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