

PATIENT INFORMATION

Hereditary Spherocytosis

What is Hereditary Spherocytosis?

As the name implies, Hereditary Spherocytosis (or HS) is an inherited disorder where the red blood cells have an abnormal shape. Normal red cells have a doughnut shape whereas people with HS have spherical red cells, which have a shortened life span due to destruction by the spleen. Haemolysis is the term applied to destruction of red cells and this can result in anaemia (a low haemoglobin concentration in the blood). Whether an individual patient with HS develops anaemia depends on the severity of haemolysis and the extent to which their bone marrow (where all blood cells are manufactured), compensates for it. Patients with fully compensated haemolysis are not anaemic and usually have a milder disease, whereas those with uncompensated haemolysis tend to have more of the problems outlined below.

How is it inherited?

HS is usually inherited in a dominant manner that affects both sexes equally. This means that both male and female children of HS patients have a 50% risk of inheriting the condition. Around 30% of patients have recessively inherited condition, which means that the risk of inheritance is much lower.

How does it affect health?

Most patients with HS have no problems with their health and do not need treatment other than supplements of a vitamin called folic acid which is necessary for red cell production of red cells by their bone marrow.

Potential problems:**Jaundice at birth**

Mild jaundice (increase in a blood pigment called bilirubin) is common at birth due to immaturity of the liver. HS increases the risk of severe jaundice in the first few days after birth. In very severe cases urgent treatment is required to reduce the level and reduce the risk of damage to the brain. If you or your partner becomes pregnant, alert the midwife and doctor in charge of the antenatal care to this risk as they may wish to make special arrangements to monitor the newborn.

Anaemia and the need for blood transfusions

Most patients with HS are not anaemic and do not need blood transfusions.

Infections

Whatever the severity of HS, all patients are at risk of developing worsening anaemia during infections, especially viral infections, such as parvovirus B19 (known as slapped cheek syndrome). This can be due to suppression of the bone marrow or increased break down of red cells. The most severe form is an “aplastic crisis” and a blood transfusion may be needed.

Symptoms include: severe fatigue, pallor or jaundice and you should seek prompt medical attention.

Gall stones

Destruction of red blood cells releases a pigment called bilirubin, which is removed from the blood by the liver. Since patients with HS have increased destruction of red cells, more bilirubin passes through the liver and gall bladder and most patients accumulate this pigment within the gall bladder. In some, this produces gallstones and inflammation of the gall bladder, which can cause problems in the late teens and beyond.

How can HS be treated?

Supportive care such as: folate supplements, blood transfusions to support acute episodes of haemolysis.

Spleen

Apart from the spherical shape, HS patients’ red blood cells function normally. Since the spleen is the site for destruction of these otherwise normal red cells, removal of the spleen (splenectomy) provides an effective cure for the condition in a majority of patients. However, splenectomy is associated with short and long-term risks that have to be weighed against the potential benefit of the procedure in the individual patient. These will be discussed with you before proceeding to splenectomy and further information about the operation and its risks will be discussed.

When to seek medical attention

If you experience:

- Sudden lethargy
- Poor appetite/feeding
- Worsening jaundice or pallor

You need to go to the Emergency Department to be reviewed

If you have any questions about the information in this leaflet, please contact:

Fiona Dooley

Haemostasis/Thrombosis Clinical Nurse Specialist

Tel: (01803) 6555270

Email: fiona.dooley@nhs.net

For further assistance or to receive this information in a different format, please contact the department which created this leaflet.