Patient Information

Royal Devon and Exeter
NHS Foundation Trust

Advice for parents of babies at risk of inheriting Hereditary Spherocytosis

What is Hereditary Spherocytosis?

Hereditary Spherocytosis (HS) is a common cause of anaemia. Anaemia means a low red cell count in the blood. The red cells in someone with HS have an abnormal covering and therefore don't hold their shape, they become sphere shaped. This means the cells are destroyed early in the body, especially in the organ the spleen.

How common is HS?

It occurs in approximately 1 in 2000 people in Northern Europe.

Is it inherited?

Yes, but not always. If a parent has HS the children each have a 50% chance of inheriting it. About ¼ of causes occur in children without a known family history, i.e. they are new cases.

What are the symptoms of HS?

Anaemia = pallor, poor energy, tiredness especially with exercise, poor growth or poor weight gain.

Jaundice = the skin looking yellow. Some infants require phototherapy (blue light treatment) in the first couple of weeks of life.

Aplastic crises = Periods of severe anaemia with viral infection, when the bone marrow production of red cells is stopped. A new onset of tiredness, pale-looking, sleeping more or not playing maybe signs of this. If a children becomes particularly unwell with a temperature or has continued symptoms of illness after the temperature has settled when advice from a doctor should be sort. Some children develop severe anaemia and require red blood cell transfusions in this illness. It is not dangerous with the right advice from doctors.

How is HS diagnosed?

HS is diagnosed by a combination of the family history, symptoms of the baby or child and blood tests.

Why don't you do the blood test sooner than 6-12 months?

If performed early in newborn babies the blood tests are misleading. The tests may falsely suggest the baby has HS when in fact they don't (false positive test result) due to the type of red blood cells newborn babies have naturally. This would led to the baby needing repeat blood tests at 6-12 months. This unclear picture may also cause more anxiety for you, the family.

Will my baby come to any harm from waiting for 6-12 months?

No, your baby won't come to harm so long as any symptoms (explained above) are acting on, if they develop.

What treatment is needed for HS?

Once the diagnosis and baseline severity are established, it is not necessary to perform repeated blood tests unless something new develops such as paleness, jaundice or tiredness. A yearly clinic appointment in the Haematology clinic will be offered and usually this is all that is needed.

Mild cases do not usually require medication or other treatment. But we have an open door policy and encourage families to represent if the child become unwell with a viral infection or abdominal pain (which may trigger investigations for gallstones).

If an aplastic crises develops, as explained above the child may require blood transfusions or treatment with steroids to increase red blood cell levels.

Moderately affected individuals are usually given medicine, a folate supplement. This helps the body produce more red blood cells. These patients may require a splenectomy. A splenectomy is an operation to remove the spleen and therefore reduce the red blood cell breakdown. This is usually performed after the age of 6 and by key hole (laparoscopic) surgery.

Severely affected children, about 3-5% of all children with HS requires regular blood transfusions. These patients are all offered a splenectomy.

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