melbourne haematology

1 fact sheet

Hereditary Spherocytosis

Overview

Hereditary Spherocytosis(HS) is a red blood cell disorder where the cells take on a shape of a ball (or sphere) instead of the normal shape of a red cell (which looks like a doughnut). Because the red cells are in the shape of a ball they are more fragile than normal red cells. The fragile red cells can break down (red cell haemolysis) and cause anaemia.

What causes HS?

HS is caused by an abnormal protein on the surface of the red cells. It is a genetic disorder so there is usually a family history of HS; HS is usually passed on from one parent to their children.

How common is HS?

HS is relatively common – around 1 person in every 5000 people has HS (around 800 people in Melbourne; population 4 million) What does having HS mean?

There are three main common problems associated with having HS

1. **Anaemia** – this occurs because the red cells break down more quickly
2. **Jaundice** – when red cells break down they release a pigment called bilirubin which appears as a yellow colour in the skin and the eyes. Over a long period of time, the problem of jaundice can be associated with gall stones.
3. **Increase in the size of the spleen** – this is the site of the red cells breaking down. The spleen is a blood filter and some of the red cells can get caught up in the spleen.

There is another uncommon potential problem for people who have HS associated with a viral infection called “Slapped cheek”. Slapped cheek is caused by a virus called Parvovirus which can infect bone marrow cells and put the red cell producing cells “to sleep”. Because patients with HS need the bone marrow to be rapidly replacing the fragile red cells, this infection can cause a severe anaemia; patients can become very pale and sometimes even need blood transfusions. Fortunately this problem is uncommon.

How is HS diagnosed?

HS is now diagnosed with a simple blood test. It is also important for a doctor to examine patients with HS to see if they are jaundiced or have an increase in the size of the spleen.

What do I need to do now that I (or my child) has been diagnosed with HS?

Most patients with HS do not need to do much about their condition at all. The bone marrow has the capacity to increase the number of red cells it produces many fold and is able to keep up replacing the fragile cells.

A few things may be helpful:

1. Regular blood tests to check on the level of haemolysis
2. Folate supplementation.
3. An important vitamin necessary for the bone marrow to function properly is folate.
   Folate is found in green leafy vegetables. Most people get enough folate in their diet but getting extra folate (particularly in HS if there is significant haemolysis – red cell breakdown) is generally recommended.
4. Ultrasounds to exclude gall stones.
5. It may also be important to make sure children and adults are not developing gall stones
   – an ultrasound of the gall bladder after that age of 5 years of age every 3 – 5 years there after, is the current recommendation.

Will I (or my child) need a splenectomy?

Removing the spleen has been used a lot in past in patients with HS; it stops the red cells from breaking down and solves the problems of HS.

Removing the spleen has problems however – the spleen is an important organ in the immune system and patients who do not have a spleen may be prone to getting serious infections. This risk is higher in younger children but may be less than previously expected because of newer vaccinations. It is now generally recommended to avoid splenectomy in children younger than 6 years and to ensure that children who may need a splenectomy have all the appropriate vaccinations.

Another approach for some of these young patients is to remove only a small part of the spleen – partial splenectomy. This is done in a few hospitals and there is some experience to say this may be very helpful for some patients.

Resources used to produce this information sheet.

- Bolton Maggs, PHB Stevens, RF Dodd, NJ Lamont, G Tittensor, P and King, M-J on behalf of the Haematology Task Force of the BCSH. *Guidelines for the diagnosis and management of hereditary spherocytosis.* BJH 2004 126 pp 455 - 474

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FURTHER QUESTIONS?

The information presented in this fact sheet is intended as a general guide only.

Patients should seek further advice and information about **hereditary spherocytosis** and their individual condition from their treating haematologist or doctor.

For additional information about blood disorders and their treatment, or to contact one of our specialist haematologists, visit the Melbourne Haematology website: **www.melbournehaematology.com.au**

