melbourne haematology

fact sheet

# Hereditary Spherocytosis (HS)

## What is Hereditary Spherocytosis (HS)?

HS is a red blood cell disorder where the cells take on the shape of a ball (sphere) instead of the normal doughnut shape of a red cell. Because the cells are ball-shaped, they are more fragile than normal red cells. These fragile 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 — HS is typically passed on from a parent to their children.

## How common is HS?

HS is relatively common — around 1 person in every 5000 has HS (around 800 people in Melbourne, population 4 million).

## What does having HS mean?

There are three main problems commonly associated with HS:

- Anaemia — because red cells break down more quickly.
- Jaundice — when red cells break down they release a pigment called bilirubin, which appears as a yellow colour in the skin and eyes. Over time, jaundice can be associated with gallstones.
- Increase in spleen size — the spleen is where red cells break down, and some cells can get caught up in it.

There is another uncommon potential problem for people with HS associated with a viral infection called "slapped cheek," caused by Parvovirus, which can infect bone marrow cells and temporarily put red-cell-producing cells "to sleep." Because patients with HS rely on the bone marrow rapidly replacing fragile red cells, this infection can cause severe anaemia — patients can become very pale and sometimes need blood transfusions. Fortunately this is uncommon.

## How is HS diagnosed?

HS is now diagnosed with a simple blood test. It's also important for a doctor to examine patients for jaundice or an enlarged spleen.

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

Most patients with HS don't need to do much about their condition at all. The bone marrow can increase red cell production many-fold to keep up with replacing fragile cells. A few things may be helpful:

- Regular blood tests to check on the level of haemolysis.
- Folate supplementation — folate is an important vitamin for bone marrow function, found in green leafy vegetables. Extra folate is generally recommended, particularly if there is significant haemolysis.
- Ultrasounds to exclude gallstones — an ultrasound of the gall bladder after age 5, then every 3–5 years, is currently recommended.

## Will I (or my child) need a splenectomy?

Removing the spleen has been used a lot in the past for patients with HS — it stops red cells breaking down and solves the problems of HS. However, removing the spleen has downsides too: it's an important immune organ, and patients without a spleen may be prone to serious infections. This risk is higher in younger children but may be less than previously expected due to newer vaccinations. It's now generally recommended to avoid splenectomy in children younger than 6 years and to ensure appropriate vaccinations for anyone who may need the procedure.

Another approach for some younger patients is to remove only part of the spleen (partial splenectomy) — done in a few hospitals, with some evidence this may help certain patients.

## References

1. Bolton-Maggs PHB, Stevens RF, Dodd NJ, Lamont G, Tittensor P, King MJ, on behalf of the Haematology Task Force of the BCSH. Guidelines for the diagnosis and management of hereditary spherocytosis. British Journal of Haematology 2004;126:455-474.

### 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 (HS)** 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**