

# Hereditary Spherocytosis

## What is Hereditary Spherocytosis (HS)?

Hereditary Spherocytosis is an inherited disorder that affects red blood cells. Normal red blood cells are do-nut shaped which enables them to change their shape to pass through small blood vessels. In Hereditary Spherocytosis the red cells are spherical and thus lack flexibility and are more prone to break when placed under physical stress. The break down of red cells can result in anaemia and / or jaundice.

The bone marrow works harder than normal to make red cells and prevent anaemia. More immature cells (reticulocytes) are present in the circulation. Rapid red cell breakdown releases haemoglobin and this can lead to jaundice, dark “coke” coloured urine and even gallstones.

Hereditary Spherocytosis is very common in people of northern European descent. A person with this condition has a 50% chance of passing this gene onto his/her children. An estimated 25% of cases are new mutations.

## What are the signs and symptoms?

- Anaemia – paleness, extreme tiredness, shortness of breath
- Jaundice - yellowing of skin and/or whites of eyes.
- Enlarged spleen (can be felt on left side of abdomen just beneath rib cage.)
- General weakness
- Irritability in children
- Abdominal pain – from splenomegaly or gallstones
- Poor growth
- Reduced energy levels and concentration
- Gall stones

## What tests are needed?

- Physical examination
- Blood tests – FBC (full blood count), reticulocyte count and liver function tests, often taken via a finger prick.

### **What treatments are available and what are their side effects?**

Most often no treatment is required.

- Splenectomy – this is the removal of the spleen. It is usually withheld if possible until the child is older ie 5 years, as the spleen is involved in helping fight common childhood infections. Splenectomy offers immediate control of the anaemia. The abnormal cell defect persists but the red cell life span returns to normal. After a splenectomy there is an increased risk of infections.
- Prophylactic (preventative) antibiotics.
- Blood transfusions. Side effects can include allergic reaction and contamination of the blood product, which occurs in less than 1 in a million. (Refer to Section 3 “Information on Blood” for further information).
- Folic acid supplementation may be recommended.

### **Problems associated with Spherocytosis**

- Aplastic Crisis - viral infections may suppress bone marrow function. Continuous red cell breakdown along with inadequate marrow production of red cells results in profound anaemia. This often requires a blood transfusion to correct.
- Haemolytic crisis - rapid red cell destruction leads to jaundice, dark urine and anaemia, which may require a blood transfusion to correct.

However it is important to note that these are usually short-lived problems.

Your child can usually live an active normal life.

## References

<http://www.medterms.com/script/main/art.asp?articlekey=3724>

<http://www.mc.vanderbilt.edu/peds/pidl/hemeonc/spherocy.htm>

<http://en.wikipedia.org/wiki/Spherocytosis>

<http://www.drhull.com/EncyMaster/S/sperocytosis.html>

Please refer to section 7 “Sources of More Information” or check the parent’s library in McGuiness & McDermott children’s clinic for further information.