Hereditary Spherocytosis (HS) – information for patients and parents

What is it HS?

HS is an inherited condition of red blood cells. Due to an inborn 'error' in the code of genes that make the surface membrane of red blood cells, it causes them to become a different shape. Instead of the red blood cells being a disc shape like normal, in HS they become round and spherical. This makes these red blood cells more fragile, so they get destroyed too quickly. Instead of surviving 3 months, they survive only a few weeks. The faster destruction of these blood cells leads to anaemia, a condition which means there is not enough red blood cells circulating in the blood.

How and when is HS diagnosed?

HS can be diagnosed at any age. In mild cases, HS may not be diagnosed until adulthood. However, if the symptoms are more severe or there is known family history of the condition, then patients are often diagnosed early in infancy.

Specialist doctors can test for the condition by performing genetic and chemical tests on blood samples. Checking the spleen can also be useful for diagnosis. The spleen is an organ in the abdomen which breaks down red blood cells. The spleen often becomes enlarged in HS, so doctors will often assess the size of this organ to help with diagnosis.

What are the symptoms of HS?

For many patients, HS is mild and there are few symptoms. However, some people can be affected more severely.

The main symptom is anaemia.

This can make patients feel tired and weak, lacking energy. They may present with jaundice- the whites of their eyes have a yellow tinge. This is not harmful but it shows that the blood cells are being destroyed too quickly. Patients may also experience:

- Lack of appetite
- Shortness of breath
- Abdominal pain – This can be caused by the spleen becoming enlarged. Round red blood cells in HS are less flexible, so it can mean they get stuck/build up in small vessels in the spleen. This can be painful.

If patients with HS become ill with a fever or a cold, it can cause their anaemia to become worse. This may mean they develop more severe symptoms, for example increased fatigue or worsened jaundice.

What are the complications of HS?

There can be some complications which may occur as a result of HS. Gallstones are small stones which can form in the gall bladder due to the increased breakdown of red blood cells. The gallbladder may need to be removed in a minor operation if it begins to cause problems or becomes painful. Sometimes, people with HS can develop severe anaemia which can require blood transfusions. If the severe anaemia persists, doctors may assess whether it would be beneficial to operate and remove the spleen so that less red blood cells are broken down (this is discussed more in the treatment section).

Aplastic crisis can be a serious complication of HS. It's usually caused when someone catches a viral infection called parvovirus. The virus causes the bone marrow to be suppressed. This is where blood cells are made, meaning that the production of red blood cells is reduced. With the decreased amount of blood cells being supplied into the blood circulation, patients can quickly develop severe anaemia.

Haemolytic crisis is the most common type of crisis in HS. Again, it is often triggered by a viral infection. It causes there to be a sudden increase in the destruction of red blood cells. The break-down of red blood cells is called haemolysis, hence the name haemolytic anaemia.

Both types of crisis can be noticed by a rapid worsening of symptoms, for example a fast decrease in energy or appetite or an increased shortness of breath. Some emergency signs to look out for include chest pain or changes in alertness. Patients experiencing aplastic or haemolytic crisis will likely need a blood transfusion.

What is the treatment for HS?

Having HS does not mean giving up on living a full and active life.

There is currently no cure for HS, however there are methods used to help reduce the symptoms it creates. Blood transfusions can be used to treat the anaemia if it becomes severe. Transfusions help to increase the number of healthy red blood cells in the blood, which can improve the lack of energy and fatigue which can be experienced. Folic acid supplements can be given to children (under the age of 5), as this is a crucial vitamin to help their body to produce more red blood cells naturally. Vitamin D supplements are also good to keep the bones strong.

More treatment options

In severe cases of HS, the spleen may need to be removed. Normally, the spleen is an organ which breaks down blood cells when they are no longer needed. In HS, it can be beneficial to remove it to help red blood cells last for longer.

The spleen also helps to produce immune cells in the body which naturally fight off infections. Patients who have had their spleen removed should then be prescribed daily preventative antibiotics and keep up to date with vaccinations to prevent these severe infections.

What are some things I can do to help?

Plenty of people want to know- is there anything I can do to help myself/my child be as well as possible with HS? While there isn't any one thing that will make all the difference, keeping well and healthy will help in the long term. This includes a good diet, staying hydrated and regular exercise, as well as staying on top of any medications prescribed e.g folic acid, vitamin D and antibiotics.

Can we help?

CAN (congenital anaemia network) is a charity specifically for patients with inherited anaemias. It was started by a group of patients and doctors, working together to tackle some of the problems of having an inherited anaemia- the lack of information and the loneliness of not knowing anyone else with your condition.

We run events for patients to meet each other, as well as provide some additional information, support and services for patients who need that extra bit of help. If you want to know more visit www.togetherwecan.uk or email us on info@togetherwecan.uk

Yes We Can

CONGENITAL ANAEMIA NETWORK