Hereditary Pyropoikilocytosis (HPP) – information for patients and parents

What is HPP?

HPP is a rare inherited disorder, in which there is an error in a gene which provides instructions for the structure of red blood cells. This error makes red blood cells more sensitive to heat, so that the membrane breaks down more easily. Therefore, red blood cells are destroyed more quickly than normal, instead of surviving 3 months, they survive only a few weeks.

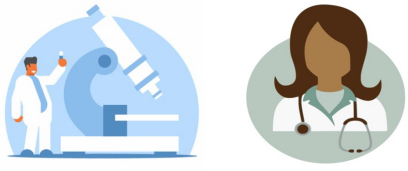
What are the basic genetics of HPP?

As mentioned before, HPP is caused by an error in a gene. Scientists often refer to errors in genes as mutations. The specific gene which is affected in HPP is called alphaspectrin. Alphaspectrin usually provides instructions to red blood cells about how to form their structure and membrane. If this process is disrupted, it causes components in the red blood cell to be made differently. This can make the red blood cells less suited to their environment, in this case they can no longer cope with the temperature of the blood.

HPP is an autosomal recessive disorder. This is the scientific term given to a condition where you must inherit two copies of the mutated gene to be affected, one from your mother and one from your father.

This means that if someone has HPP, they have two copies of the mutated alphaspectrin gene. It's also possible to be a carrier of HPP. This is someone who has only inherited one copy of the mutated gene. Therefor, they do not have HPP themselves, but they are able to pass the mutated alphaspectrin gene on to their offspring.

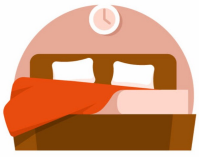
How and when is HPP diagnosed?



Diagnosis usually happens in early childhood.

Since the membrane and structure of red blood cells is affected in HPP, it causes them to be a different shape compared to normal. This means that the condition can be spotted if a doctor looks at the shape of patients red blood cells. This can also be referred to as checking the morphology (shape or structure) of the red blood cells. A sample of the patient's blood will be taken and looked at under a microscope. The doctor can also perform some chemical tests on the blood sample to understand if the patient has HPP or not.

What are the symptoms of HPP?



Symptoms can be different for each person, they can vary from mild to severe.

The main symptom of HPP is haemolytic anaemia. Haemolysis is the name for the process of breaking down red blood cells, and anaemia is when there is not enough red blood cells in the blood. Haemolytic anaemia is anaemia caused by red blood cells being destroyed too quickly.

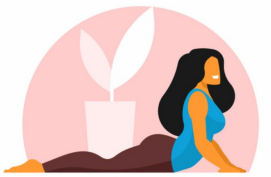
Often, haemolytic anaemia can be severe in HPP. This can present from soon after birth and cause babies to be jaundiced. This is when their skin and the whites of their eyes becomes a yellow colour. Haemolytic anaemia can cause patients to be fatigued, lacking energy and weak. Their skin might be pale or jaundiced and they may experience confusion, dizziness and a decreased ability to concentrate.

Some other symptoms of HPP can include:

- Gallstones - These 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.

- Enlarged spleen – The spleen is an organ is the stomach which is responsible for the break down of the blood cells. If there is increased haemolysis (break down of red blood cells), the spleen is working extra hard and therefor it can increase in size. This can sometimes cause abdominal pain or discomfort. In serious cases of haemolytic anaemia, the spleen can be removed in an operation, to help red blood cells to last for longer.

What is the treatment for HPP?



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

Although there is not a cure for HPP, there are ways to improve symptoms. Blood transfusions can be given to treat the haemolytic anaemia which is experienced in HPP. Transfusions increase the number of healthy red blood cells circulating in the blood, helping to relieve the symptoms of fatigue and lack of energy.

Your doctor may also suggest taking folic acid supplements. This is a good idea as this vitamin is used in the production of red blood cells. It can help to produce more red blood cells, which can replace the one which are destroyed in the haemolytic anaemia. This can also improve symptoms.

What can I do to help myself/my child with HPP?

Staying generally well is an important way to help manage HPP. Making sure you are hydrated, have a balanced diet and doing regular exercise will all contribute to maintaining health.

For people with HPP, becoming unwell can cause worsened anaemia and therefor increased symptoms like fatigue and weakness. This is why it's advised to stay hygienic and be up to date with vaccinations, as this will help to prevent illness and colds.

If your child has been diagnosed with HPP, you may notice they get more tired than other children their age. For example, they may need a nap after school, they might get more worn out in physical activities or struggle with their concentration in lessons. It could be a good idea to make their teachers aware of your child's condition, so they can have breaks or extra help if they need it.

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