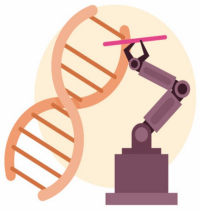
Haemoglobin H disease (HbH disease)

What is HbH disease?

HbH disease is a form of alpha thalassemia. Alpha thalassemia is an inherited condition in which reduced amounts of alpha globin are produced. Alpha globin is a building block of haemoglobin, the part of a red blood cell that transports oxygen around in the blood. This causes there to be less haemoglobin present in the blood and therefor less oxygen can be delivered to tissues of the body.



What causes HbH disease?

HbH disease is an inherited condition, meaning that it's passed down through families. HbH disease is caused by an error in a gene, which is referred to as a mutation in scientific terms. It's an autosomal recessive condition which means both the mother and father must carry the mutation in order to pass it on to their child.

In HbH disease, the mutations affect genes which provide instruction for the production of apha globin. Since alpha globin is a building block of haemoglobin, there is disruption to the production of haemoglobin. This is the cause for the reduced amounts of haemoglobin and therefore red blood cells. There are many genes which are involved in this process, so mutations in multiple different genes could be responsible.

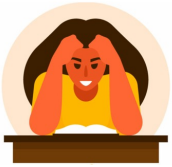
How and when is HbH disease diagnosed?

HbH disease is diagnosed via genetic testing. Doctors will take a sample of blood and use this to look at the genetics of the individual. They can perform some chemical tests on the sample to identify whether the genetic mutation is present in the persons DNA. From here, doctors can understand whether the individual is affected with HbH disease or not. They can also identify if someone is a carrier of the condition. This means they do not present with the condition themselves, however they have the potential to pass it on to their offspring.

If someone in your family has previously been diagnosed with HbH disease, it is possible for a baby to be tested for HbH disease whilst the mother is still pregnant. This is called prenatal testing.

HbH disease should be considered in babies that are born with a specific type of anaemia, microcytic haemolytic anaemia. Diagnosis often happens at birth or in early childhood, however some people may be diagnosed at later stages.

What are the symptoms of HbH disease?

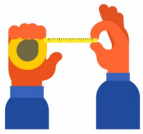


Symptoms can vary from mild to severe depending on each person.

Patients with Hbh disease usually experience moderate to severe anaemia. Anaemia is when there is a reduced number of red blood cells circulating in the blood.

Anaemia can cause patients to be fatigued, lacking energy and weak. Their skin might be jaundiced (a yellow colour) and they may experience confusion, dizziness and a decreased ability to concentrate.

Some people can experience shortness of breath, a fast heart rate and also moodiness or irritability.



Children with HbH disease may have stunted growth and development. This is due to the condition causing reduced amounts of growth hormone to be produced. It can mean that these children are shorter and skinnier than others their age. In some cases, there can also be a change in the shape of bones in the face and the head.

A complication of HbH disease can be a build up of iron in the body. This can be caused by the disease itself or from blood transfusions which are sometimes used in treatment. Excess iron can be removed by a method called iron chelation. It's important to remove it, otherwise a build up of iron can damage the heart, liver and the hormone system. Some people may also experience an enlarged spleen or gallstones, both of which can be treated in minor operations if they become painful or start to cause problems.

What is the treatment for HbH disease?

HbH disease in an inherited condition so there is no cure, however there are ways to manage the symptoms. Patients can be treated with blood transfusions, depending on the severity of their condition. This helps to increase the number of healthy red blood cells which are circulating in the blood and therefor reduce the symptoms like tiredness and lacking energy. In milder cases, patients often only require occasional blood transfusions. However, regular transfusions might be needed if someone is affected more severely.

Patients with HbH disease will often have ongoing monitoring of blood counts, growth, bone health, levels of fatigue and also the size of the spleen. This helps doctors to understand the severity of each case, which means they can intervene with blood transfusions and treatment if they need to.

It is recommended that patients experiencing anaemia are supplemented with folic acid. This is a vitamin which is essential in the production of red blood cells.

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

Being generally healthy is always a good way to maintain good well being whilst also helping to prevent diseases becoming worse. This can be done by staying hydrated and having a balanced diet, along with regular exercise and physical activities. Smoking and drinking excess alcohol can worsen anaemia, so avoiding this is also good.

It's important to make sure that you or your child are in contact with your doctors and are getting regular medical care. This will help to keep on top of treatment and monitoring, which will overall improve your/your childs well being and symptoms.

CAN (congenital anaemia network) is a charity specifically for patients with inherited anaemias. 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