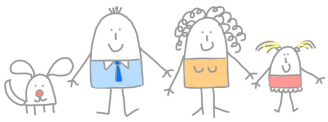
Thalassemia intermedia

What is it?

Thalassemia intermedia is a subgroup of beta thalassemia. Beta thalassemia is an inherited blood disorder in which there is a problem with the production of beta globin. Beta globin is a building block of haemoglobin, the part of the red blood cells that transports oxygen around in the blood. Reduced beta globin means reduced haemoglobin, which further means less oxygen is being delivered to tissues of the body.

Thalassemia intermedia is the condition that encompasses cases of beta thalassemia which are of medium severity, not mild but also not severe.

What is the cause?



Thalassemia intermedia is an inherited condition, so it is caused by a genetic error being passed down through families. Errors in genes are referred to as mutations in scientific terms. It is an autosomal recessive disorder. This means that there must be two copies of the mutation gene inherited, one from the mother and one from the father, in order to be affected with thalassemia intermedia.

How and when is it diagnosed?

Thalassemia intermedia can be tested for by taking a blood sample. Doctors will then look at the blood to check the size, number and maturity of the red blood cells. They can also assess the amount of haemoglobin in the sample and perform some chemical tests to look at the DNA. All of this will allow doctors to identify if an individual has thalassemia intermedia.

It can be diagnosed in children as young as age two – they may present with growth and development problems as a cause of thalassemia intermedia. In other people with milder thalassemia intermedia, they may not present with symptoms until adulthood.

What are the symptoms?

The main symptom is moderate to severe anaemia. Anaemia is when there is reduced amounts of red blood cells circulating in the blood.

The anaemia experienced can give rise to a variety of symptoms. This includes extreme tiredness, a lack of energy and weakness. It can also cause headaches, dizziness and make it hard to concentrate. Often, people have paleness and some people might notice that their skin in jaundiced (a yellow colour). In some cases, thalassemia intermedia can cause delayed growth and development, which may mean that children with this condition are shorter and thinner than other children their age. It can also cause bones to be weaker.

What is the treatment?

Thalassemia intermedia is a hereditary condition so there is no cure, however there are methods which are used to improve the symptoms. Although patients do not usually require regular blood transfusion, this is an option if their anaemia worsens.

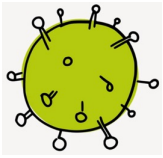
Thalassemia major is the more severe form of beta thalassemia, in which patients are dependant on regular blood transfusions.

Patients will be monitored to make sure that levels of iron in their body don't become too high. This can happen due to thalassemia intermedia itself or as a result of blood transfusions. Iron chelation therapy is the process used to remove excess iron, to prevent it from building up and damaging the organs.

It is recommended that people experiencing anaemia should be supplemented with folic acid daily. This is a vitamin which is needed for the production of red blood cells, so it's important to have enough. Sometimes anaemia can cause complications like an enlarged spleen or gallstones. Both of these can be treated with operations if they become painful or begin to cause problems.

What can you do to help?

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 is important to avoid infections or becoming ill whenever possible as this can cause anaemia to get worse. This can be done by staying up to date with vaccinations and maintaining good hygiene and hand washing.

Make sure you are keeping in regular contact with your doctors. This will help to monitor your condition and spot early signs if things start to get worse. Also, remember to reach out for support if you need it.

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