Chiesi
global rare diseases

OUR IRON WILL

not wear us out.
not overload our organs.
not threaten our future.

THANKS TO OUR IRON WILL

UNDERSTANDING THALASSAEMIA

SELECT AN OPTION TO GET STARTED >

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Date of preparation March 2023 | ALL_23_216

WHAT IS THALASSAEMIA?

1

There are multiple forms of thalassaemia with varying severity[1]

Thalassaemia, a group of conditions that affects a substance in the blood called haemoglobin, is among the most common inherited disorders in the world[1–3]

There are two main types:[1,4,5]

- **Alpha thalassaemia**
  (caused by having faulty alpha globin genes)

- **Beta thalassaemia**
  (caused by having faulty beta globin genes)

Beta thalassaemia major is the most severe form of the disease, and requires long-term blood transfusions to manage symptoms[1,6,7]

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WHAT IS THALASSAEMIA?

1

Thalassaemia is associated with anaemia[1]

The main health conditions associated with thalassaemia are:[1]

Anaemia, which can cause severe tiredness, weakness, shortness of breath, heartbeat irregularities and pale skin

Too much iron in the body, caused by the regular blood transfusions used to treat anaemia – if unmanaged, excess iron can cause problems with the heart, liver and other organs

Delayed growth, weak and fragile bones (osteoporosis) and reduced fertility can occur in some people

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WHAT IS THALASSAEMIA?

1

Signs of thalassaemia can present during early childhood[1,8]

Individuals with more severe forms of thalassaemia usually have conditions such as anaemia that come to medical attention within the first two years of life.[8]

Without treatment, children with severe forms of thalassaemia have severe failure to thrive and shortened life expectancy[8]

However, **appropriate treatment allows for normal growth and development[8]**

Individuals with milder forms of thalassaemia may present with symptoms later in life.[1]

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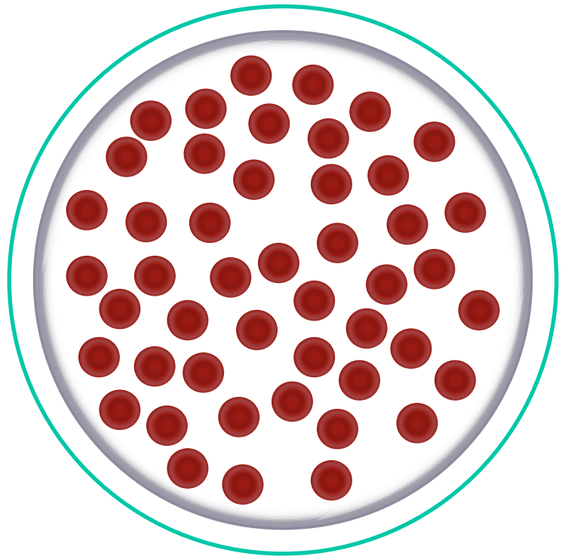
WHAT CAUSES THALASSAEMIA?

Thalassaemia is an inherited disorder caused by having faulty genes important for haemoglobin[1,9]

Thalassaemia is caused by having faulty alpha globin genes (in alpha thalassaemia) or beta globin genes (in beta thalassaemia).[1,4,5,9]

The faulty genes affect the production of a substance in red blood cells called haemoglobin, reducing the cells' ability to carry oxygen – leading to anaemia.[1,10]



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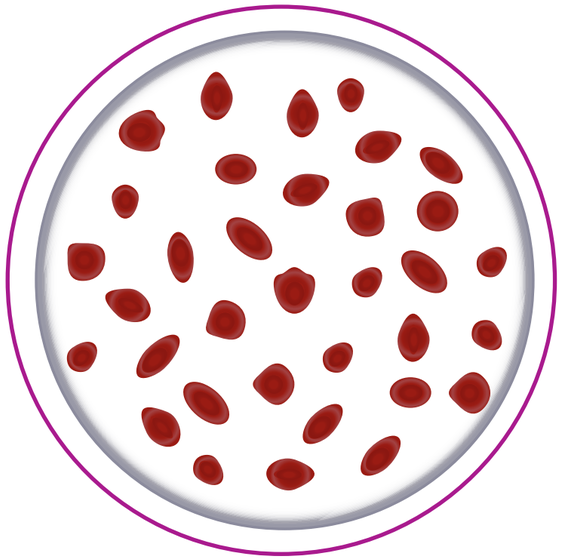
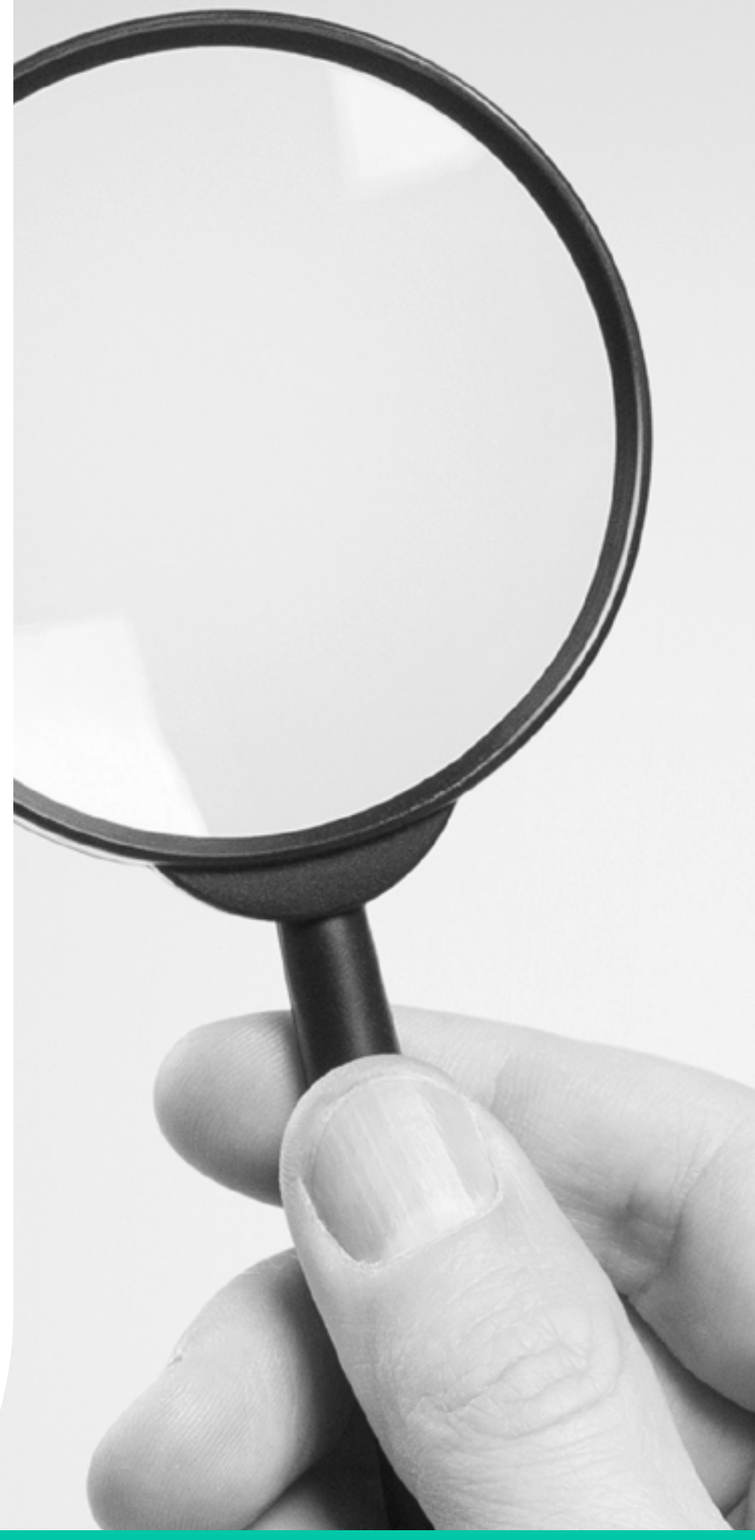


Affected

Reduced oxygen carrying ability[10]



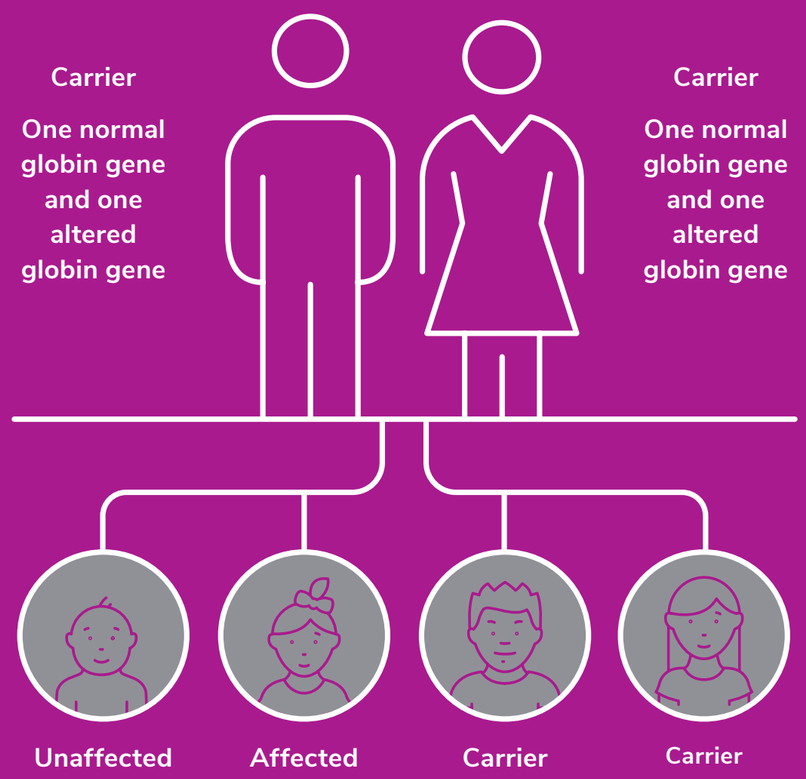
WHAT CAUSES THALASSAEMIA?

2

People can inherit thalassaemia from their parents – and can pass on the condition to their children[8,9]

If a person inherits faulty globin genes from their parents, they may develop the disease.[8,9]

Example inheritance pattern (beta thalassaemia)[9]



TREATMENT OF THALASSAEMIA[11]

3

Many people with thalassaemia will require regular blood transfusions

Most people with severe forms of thalassaemia will need to have regular blood transfusions to treat the anaemia associated with the condition.

Transfusions involve being given blood through a tube inserted into a vein in your arm; how often they are needed depends on the severity and type of thalassaemia.

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UNDERSTANDING IRON OVERLOAD[12]

However, blood transfusions can lead to the accumulation of toxic quantities of iron...

Because the body lacks the ability to get rid of excess iron, if too much is taken in, **iron can circulate in the blood and form deposits in tissues and organs** – where it causes damage.

Blood transfusions contain iron. This is why people with thalassaemia receiving them can have too much iron in their bodies, risking damage to their:

- Liver
- Thyroid and other endocrine glands
- Heart
- Pancreas

Iron overload affecting the heart is the leading cause of mortality among people with thalassaemia major[13,14]

TURN OVER

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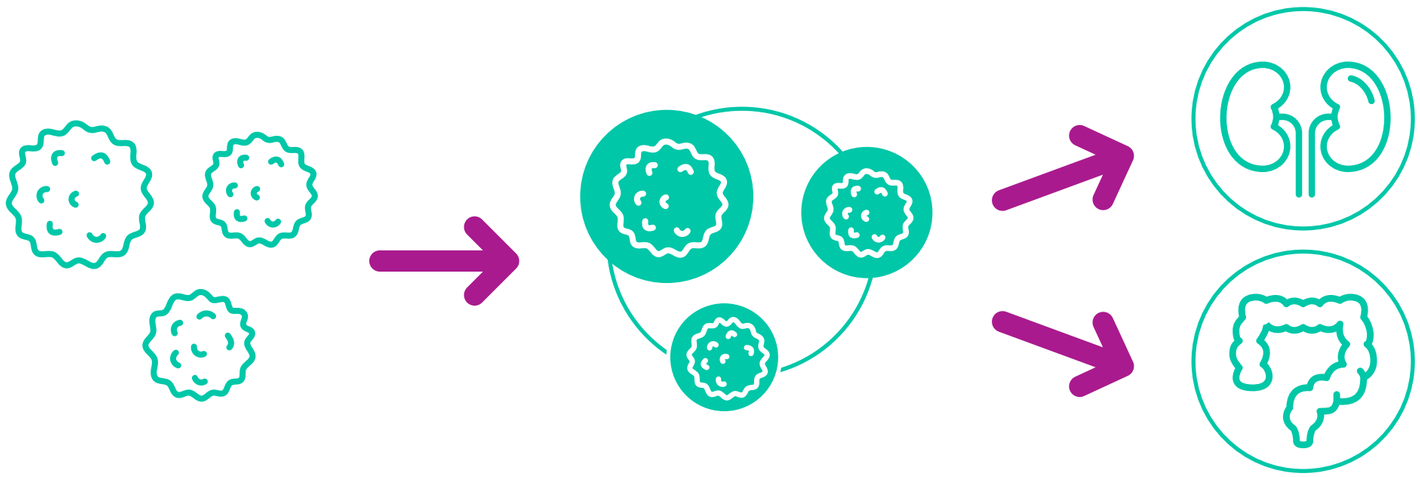
UNDERSTANDING IRON OVERLOAD[15]

...which is why transfusions are accompanied by a therapy that removes excess iron

The extra, toxic iron is removed from the body using a form of treatment called 'iron chelation'. These iron chelators bind the free iron in the body and allow the body to excrete it.



Toxic iron builds up in the body

Toxic iron is bound by iron chelation therapies

The bound iron can then pass out of the body in urine or faeces

When iron is removed from the heart, heart function improves and the risk of heart failure decreases[16]

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WHAT DOES IT MEAN FOR ME?

5

When properly managed, many people can live a near-normal life with thalassaemia[17]

1. Get regular check-ups and follow your doctor's advice[18,19]
2. Learn healthy habits[18–20]
3. Manage your infection risk[18,19]
4. Patient support groups can help[21,22]

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WHAT DOES IT MEAN FOR ME?

5

When properly managed, many people can live a near-normal life with thalassaemia[17]

1 Get regular check-ups and follow your doctor's advice[18,19]

Depending on how severe your thalassaemia is, you might need to receive regular blood transfusions.[1,11]

This will usually be accompanied by treatment to prevent iron overload, which can be a consequence of your transfusions.[1,11]

If you receive regular blood transfusions, your doctor may monitor for iron overload in your body and tailor your iron chelation therapy to your needs. It is very important to follow your doctor's prescription to ensure iron levels stay where they should be – to help avoid complications.[23–25]

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WHAT DOES IT MEAN FOR ME?

When properly managed, many people can live a near-normal life with thalassaemia[17]

2 Learn healthy habits[18–20]

A well-balanced and healthy diet (also including dietary supplements where necessary) can improve your overall health.[18–20]

Try and fit in regular moderate exercise to keep you healthy.[18–20]

DO: Talk to your doctor for help with your healthy lifestyle habits[18]

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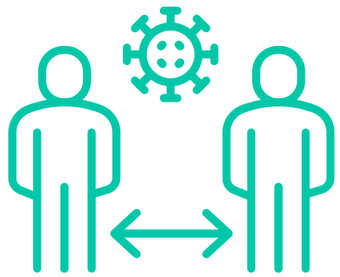
WHAT DOES IT MEAN FOR ME?

When properly managed, many people can live a near-normal life with thalassaemia[17]

3 Manage your infection risk[18,19]

Infections are important complications for people with thalassaemia. There are steps you can take to help prevent everyday infections.[18,19]

Wash your hands with soap and water regularly[19]



Avoid close contact with sick people when possible[19]

Ensure all your vaccinations are up-to-date[19]

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WHAT DOES IT MEAN FOR ME?

5

When properly managed, many people can live a near-normal life with thalassaemia[17]

4 Patient support groups can help[21,22]

Think holistically about your health: physical, mental and social wellbeing all play important roles in your wellbeing.[17]

Your healthcare team, friends, family, partners, patient support groups and others can provide information, assistance and support to help you live a healthy life. Make use of them![18–22]

UNITED KINGDOM THALASSAEMIA SOCIETY UKTS

TIF THALASSAEMIA INTERNATIONAL FEDERATION

GREEK THALASSAEMIA ASSOCIATION

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YOUR CHILD AND THALASSAEMIA

It can be difficult when faced with a diagnosis of a long-term condition for your child. However, it's important to remember that **there is support available.**

Look to your child's healthcare team, your friends and family, and support groups.

Up to 400,000 babies are born every year with a severe haemoglobin disorder like thalassaemia[1,26] – **so you are not alone.**

Remember that, with appropriate management, **many people with thalassaemia can live a near-normal life.**[17] Think about all of the opportunities your child can have in life.

Visit other sections to learn more

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USEFUL RESOURCES

7

For useful resources and more information about iron overload, iron chelation and living with thalassaemia, visit:

ourironwill.net

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SUMMARY

Thalassaemia is a group of inherited (passed on through genes) conditions that **affects a substance in the blood called haemoglobin**[1–3,8,9]

Individuals with more severe forms of thalassaemia usually have **conditions such as anaemia that come to medical attention within the first two years of life**[8]

Most people with severe forms of thalassaemia **will need to have regular blood transfusions** to treat the anaemia associated with the condition[11]

Regular blood transfusions can lead to the accumulation of toxic quantities of iron, which is why **transfusions are accompanied by a therapy that removes excess iron ('iron chelation therapy')**[12,15]

Iron overload affecting the heart is the leading cause of mortality among people with thalassaemia major.[13,14] **When iron is removed from the heart, heart function improves and the risk of heart failure decreases**[16]

When properly managed, many people can live a near-normal life with thalassaemia[17]

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