OUR IRON WILL

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

THANKS TO OUR IRON WILL

YOUR GUIDE TO:

UNDERSTANDING THALASSAEMIA

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CONTENTS

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WELCOME

ARE YOU... A parent with a diagnosis for your child?

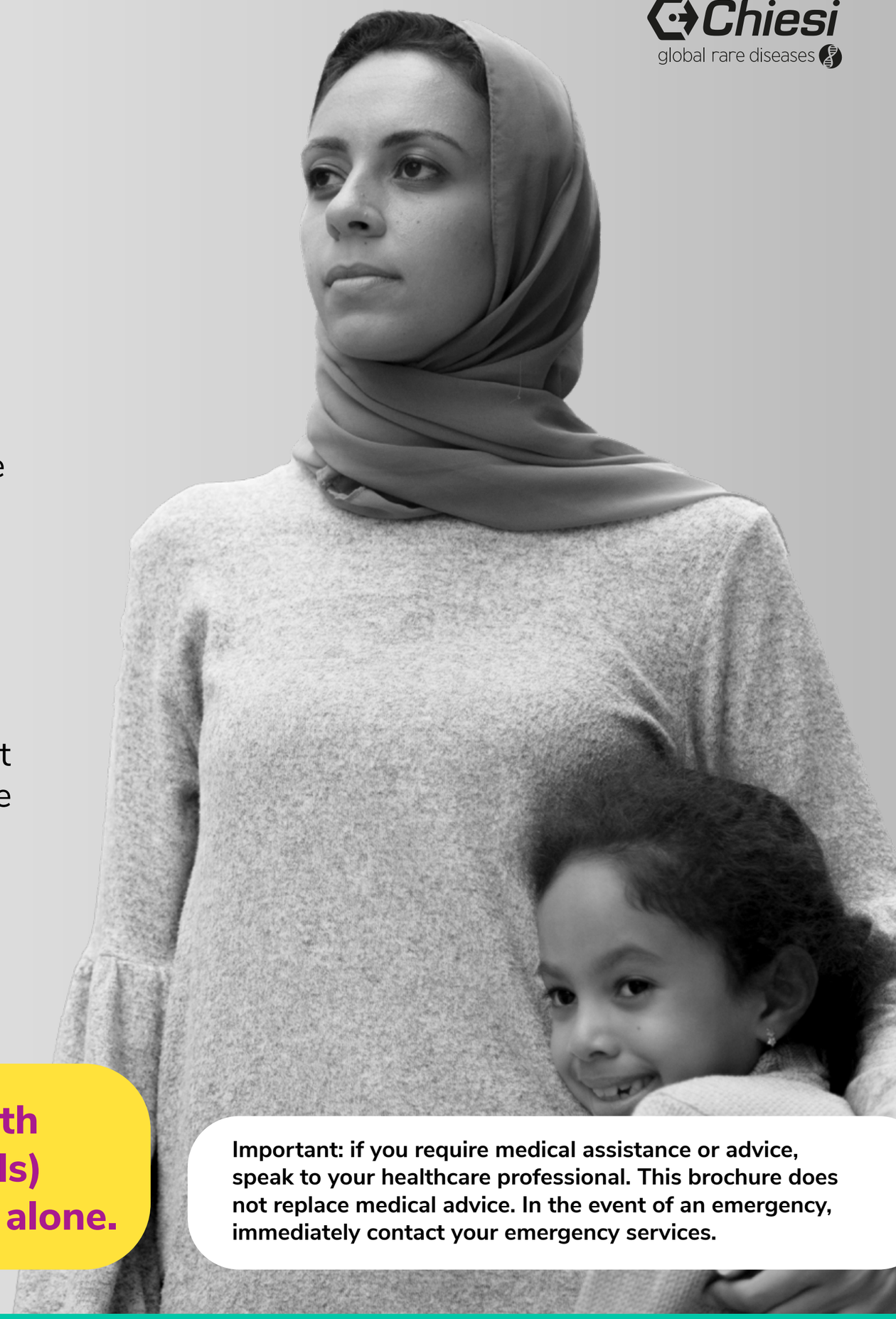
Facing a diagnosis of a long-term condition for your child can be challenging. It is important to remember that there is support available.

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

Remember that, with proper management, many people with thalassaemia can live near-normal lives.[2] Think about the opportunities your child has in life; do not dwell on the limitations you think they could face.

In this brochure, you can learn more about what to expect from your child's condition.

Up to 400,000 babies are born every year with serious haemoglobin (a part of red blood cells) disorders like thalassaemia[1] – so you are not alone.



Important: if you require medical assistance or advice, speak to your healthcare professional. This brochure does not replace medical advice. In the event of an emergency, immediately contact your emergency services.

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WELCOME

ARE YOU... Someone who has thalassaemia and wants to learn more?

Depending on the form and severity of your thalassaemia, you may have been receiving treatment since you were very young.[3,4]

Whenever you discovered that you had thalassaemia, it is important to remember that proper management of the condition means that you could live a near-normal life.[2]

In this brochure, you can find information that will help you understand your condition and learn some ways to ensure that your lifestyle is as healthy as possible.

Proper management of the condition means that you could live a near-normal life.[2]

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WHAT IS THALASSAEMIA?

Thalassaemia is a group of inherited conditions that affects haemoglobin – a substance in the blood.[3,5,6]

The conditions are inherited, which means the **genes you inherit from your parents determine whether you develop** thalassaemia.[4,7]

This also means that if you have thalassaemia or if you are a carrier of the faulty genes that cause thalassaemia, **you may pass on thalassaemia to your children.**[4,7]

Many forms exist, but there are two main types of thalassaemia:[3,7–9]

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

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

Beta thalassaemia major is the most severe form of the disease. It requires long-term blood transfusions to manage symptoms.[3,10,11]

Thalassaemia is a group of inherited conditions associated with anaemia[3,5-7,12,13]

The science

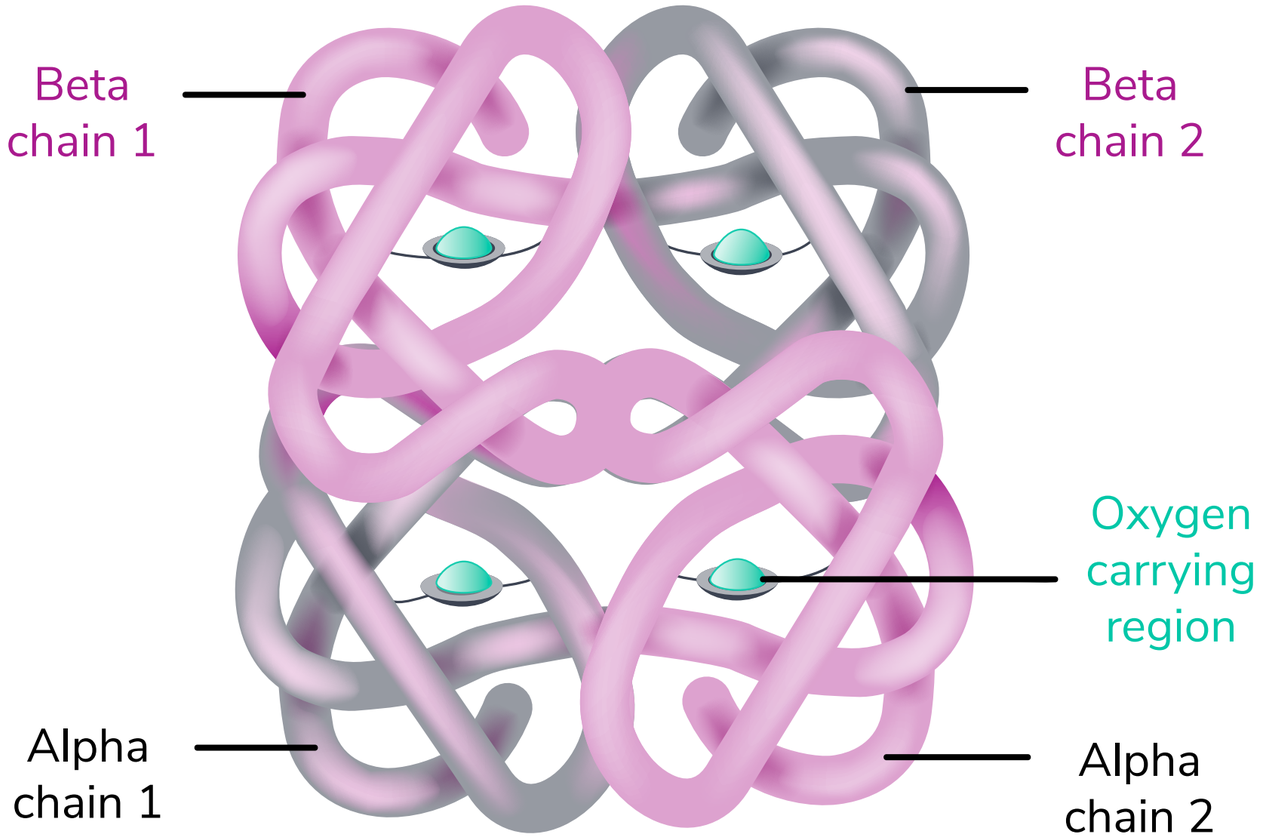
The faulty genes causing thalassaemia disrupt the structure of the oxygen-carrying substance in red blood cells called **haemoglobin**, reducing the cells' ability to carry oxygen around the body.[7.12]

This can lead to anaemia, where the tissues and organs in your body do not get enough oxygen.[13]

SEE MORE

A CLOSER LOOK

It is the alpha and beta ‘chains’ of haemoglobin that are affected in thalassaemia, leading to faulty haemoglobin and fewer healthy red blood cells[14,15]



Adult haemoglobin

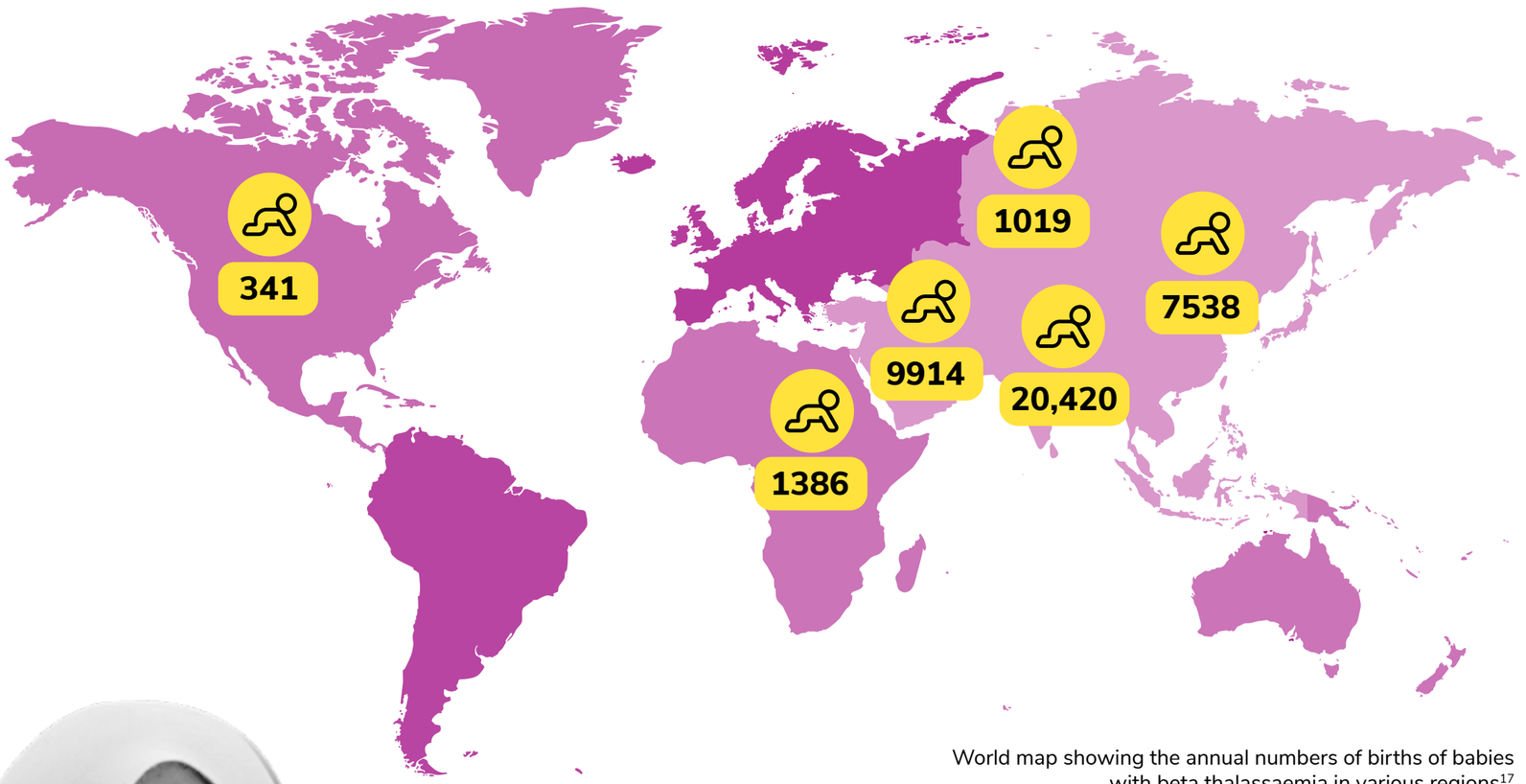
Haemaglobin is found inside red blood cells, where it carries oxygen in the blood – from the air we breathe to all the tissues and organs in our bodies.[14,15]

What is haemoglobin?

It is a substance in the blood responsible for carrying and delivering oxygen throughout the body. It gives the blood its red colour.[14–16]

Most haemoglobin in adults is made of two different parts, referred to as alpha and beta ‘globin chains’. Thalassaemia is caused by faulty genes that leads to decreased synthesis of alpha or beta globin chains, resulting in reduced production of normal haemoglobin and healthy red blood cells in the body.[15]

HOW MANY PEOPLE HAVE THALASSAEMIA?



World map showing the annual numbers of births of babies with beta thalassaemia in various regions[17]

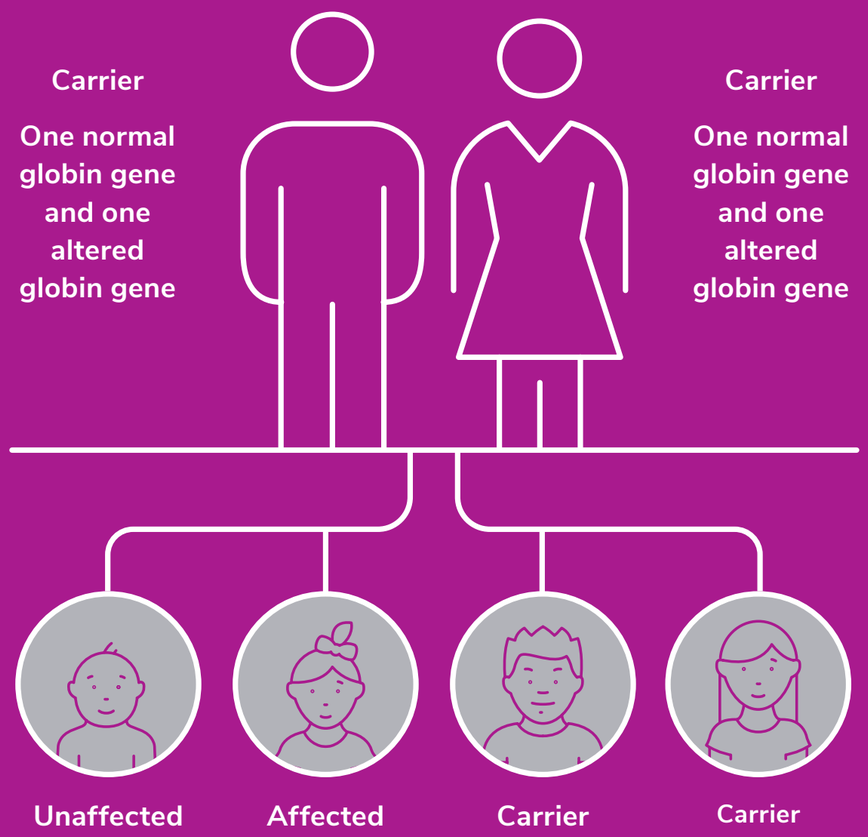
If you have thalassaemia, you are not alone. **It is among the most common groups** of inherited disorders in the world.[3,5,6,18]

Up to 400,000 babies are born every year with severe haemoglobin disorders like thalassaemia.[1]

Thalassaemia is among the most common inherited disorders[3,5,6]

INHERITANCE OF THALASSAEMIA

Example inheritance pattern (beta thalassaemia)[7]



Globally, it is estimated that there are 270 million ‘carriers’ with genes for abnormal haemoglobins and thalassaemia.[1] People with faulty genes, whether they have symptoms of thalassaemia or not, can pass on those faulty genes to their children. Whether or not the child develops thalassaemia will depend on the genes inherited.[4,7]

Remember: living with thalassaemia **does not necessarily mean that the person will develop severe disease** – many forms with different severities exist.[3,9]

The faulty genes causing thalassaemia can be inherited[4,7]

WHAT ARE THE SYMPTOMS OF THALASSAEMIA?

There are multiple forms of thalassaemia with varying severity.[3]

The main health conditions associated with thalassaemia are:[3]

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 hormones

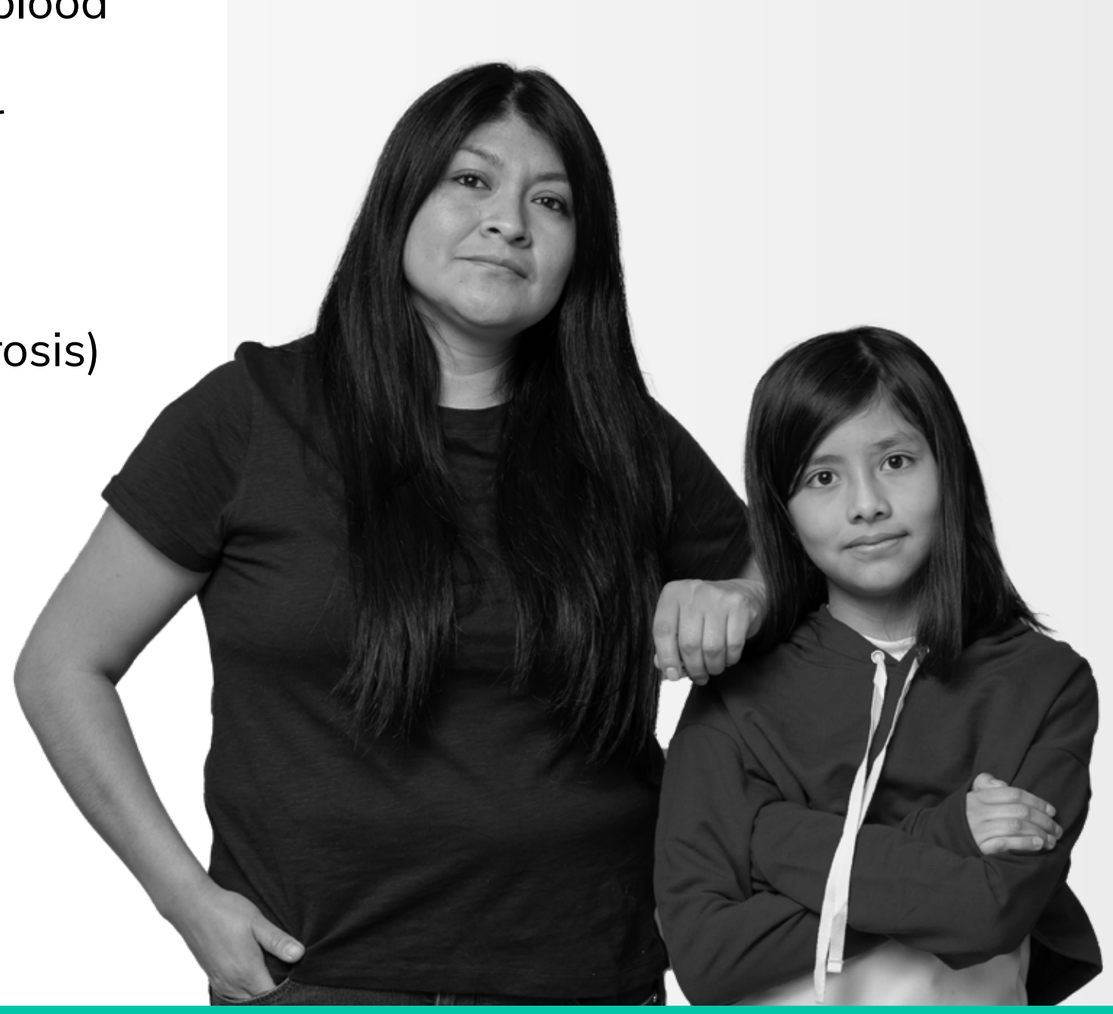
Delayed growth, weak and fragile bones (osteoporosis) and reduced fertility can occur in some people

Signs of more severe forms of thalassaemia often present during early childhood – within the first two years of life.[3,4]

Milder forms may go unnoticed until later in life.[3]

Children

Without treatment, children with severe forms of thalassaemia have severe failure to thrive and shortened life expectancy. However, **appropriate treatment allows for normal growth and development.**[4]



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HOW IS THALASSAEMIA TREATED?

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Most people with severe forms of thalassaemia will need to have **regular blood transfusions to treat the anaemia** associated with the condition. How often they are needed depends on the severity and type of thalassaemia.[19]

Various other medicines and treatments may also be used, depending on how thalassaemia affects you or your child. These could include:[19]

- Hormones to help trigger puberty in children with delayed puberty and treat low hormone levels
- Vaccinations and antibiotics to prevent and treat infections
- Thyroid hormones to treat problems with the thyroid gland
- Bisphosphonates to help strengthen the bones
- Gallbladder removal surgery to treat gallstones
- Regular monitoring to check for heart problems may also be used
- Stem cell or bone marrow transplants are occasionally used to treat thalassaemia, but this is normally rare due to the significant risks involved

Keep going forwards

Appropriate treatment allows for normal growth and development, and **allows for a near-normal life.**[2,4]

Are you a parent?

Making sure that all of your child's transfusions and iron chelation therapy sessions when instructed by your child's doctor are completed can feel like a burden – but optimal treatment of thalassaemia is important for helping someone with the condition to live a normal life.[2,4]

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UNDERSTANDING IRON OVERLOAD

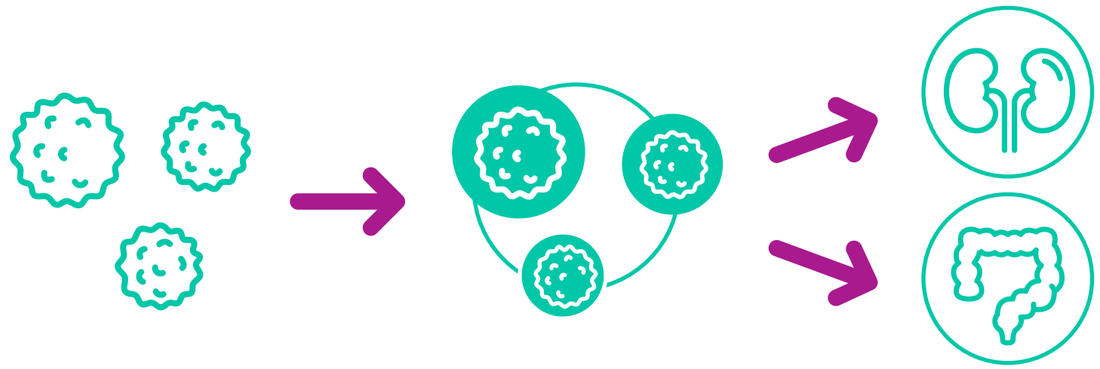
Blood transfusions can lead to toxic accumulation of iron...[20]

Because the body lacks the ability to get rid of excess iron, if too much iron is taken in, it can circulate in the blood and damage tissues and organs.[20]

Blood transfusions contain iron. This is why people with thalassaemia receiving transfusions can have too much iron in their bodies, risking damage to or impairing the function of their liver, heart, kidneys, pancreas, and thyroid and other endocrine glands.[20]

...which is why transfusions are accompanied by a therapy that removes excess iron[21]

The extra iron is removed from the body using 'iron chelation therapy'. This binds the extra iron and allows the body to excrete it:[21]



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

Iron overload and the heart

In the past, iron overload affecting the heart lead to early mortality in patients with thalassaemia major. However, with today's medicine, many of the toxic effects of iron overload on the heart can be largely prevented or reversed.[22]

Despite this improvement, it is important to note that iron overload affecting the heart remains the leading cause of mortality among people with thalassaemia major[23,24] – which is why it is important to stay on therapy to help iron chelation treatment succeed.[22,28,29] When iron is removed from the heart in this way, heart function improves and the risk of heart failure drops.[25]

WHAT CAN I DO?

1 Get regular check-ups and follow your doctor's advice[26,27]

If you receive regular transfusions, your doctor may monitor for iron overload throughout 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.[22,28,29]

2 Learn healthy habits[26,27,30]

A well-balanced diet (which can include dietary supplements where necessary) can improve overall health.[26,27,30] If appropriate, try and fit in regular moderate exercise.[26,27,30]

3 Manage your infection risk[26,27]

Infections are important complications for people with thalassaemia.
There are steps you can take to help prevent everyday infections.[26,27]

4 Patient support groups can help[31,32]

Think holistically about health: physical, mental and social wellbeing all play important roles in wellbeing.[2]

Healthcare teams, friends, family, partners, patient support groups and others can provide information, assistance, and support to help you or the person you care for to live a healthy life.[26,27,30–32]

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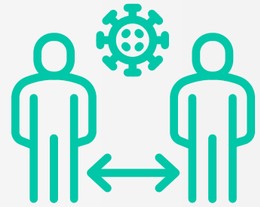
There are ways that you can stay as healthy as possible – so that you can make the most of life and seize your opportunities.

TIPS

Wash your hands with soap and water regularly[27]



Avoid close contact with sick people when possible[27]

Ensure all your vaccinations are up-to-date[27]

USEFUL LINKS

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

ourironwill.net

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REFERENCES

1. De Sanctis V, et al. *Mediterr J Hematol Infect Dis*. 2017;9(1):e2017018.
2. Angastiniotis M. LIFESTYLE AND QUALITY OF LIFE. In: Cappellini MD, Cohen A, Porter J, et al., editors. Guidelines for the Management of Transfusion Dependent Thalassaemia (TDT) [Internet]. 3rd edition. Nicosia (CY): Thalassaemia International Federation; 2014. Chapter 16. Available from: https://www.ncbi.nlm.nih.gov/books/NBK269383.
3. NHS. Overview Thalassaemia. Available at: https://www.nhs.uk/conditions/thalassaemia/#:~:text=Thalassaemia%20is%20the%20name%20for,short%20of%20breath%20and%20pale) [accessed January 2023].
4. Origa R. Beta-Thalassemia. 2000 Sep 28 [Updated 2021 Feb 4]. In: Adam MP, Everman DB, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1426/.
5. Marengo-Rowe AJ. *Proc (Bayl Univ Med Cent)*. 2007;20(1):27–31.
6. Higgs DR. *Cold Spring Harb Perspect Med*. 2013;3(1):a011718.
7. NHS. Thalassaemia – Causes. Available at: https://www.nhs.uk/conditions/thalassaemia/causes/ [accessed January 2023].
8. Mettananda S. *Front Genome Ed*. 2021;3:752278.
9. Genomics Education Programme. Alpha-thalassaemia. Available at: https://www.genomicseducation.hee.nhs.uk/documents/alpha-thalassemia/ [accessed January 2023].
10. Shah FT, et al. *Blood Rev*. 2019;37:100588.
11. Farmakis P, et al. *HemaSphere.* 2022:6(8):e732.
12. Needs T, Gonzalez-Mosquera LF, Lynch DT. Beta Thalassemia. [Updated 2022 May 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK531481/.
13. Bupa. Anaemia. Available at: https://www.bupa.co.uk/health-information/heart-blood-circulation/anaemia [accessed January 2023].
14. UCSF Benioff Children's Hospital. What is Thalassemia? Available at: https://thalassemia.com/what-is-thal-beta.aspx#gsc.tab=0 [accessed January 2023].
15. Bajwa H, Basit H. Thalassemia. [Updated 2022 Aug 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK545151/.
16. NHS Blood and Transplant. Haemoglobin and iron. Available at: https://www.blood.co.uk/the-donation-process/further-information/haemoglobin-and-iron/ [accessed January 2023].
17. Colah R, et al. *Expert Rev Hematol.* 2010;3(1):103–117.
18. Vlok M, et al. *Sci Rep*. 2021;11(1):5677.
19. NHS. Thalassaemia – Treatment. Available at: https://www.nhs.uk/conditions/thalassaemia/treatment/ [accessed January 2023].
20. Shander A, et al. *Vox Sang.* 2009;97(3):185–197.
21. Kushner JP et al. *Haematology.* 2001;1:47–61.
22. Coates TD. *Hematology Am Soc Hematol Educ Program*. 2019;2019(1):337–344.
23. Lekawanvijit S, Chattipakorn N. Can *J Cardiol.* 2009;25(4):213–218.
24. Meloni A, et al. *Blood*. 2022;140(Supplement 1):5369–5370.
25. Pennell DJ, et al. *J Cardiovasc Magn Reson*. 2011;13(1):45.
26. CDC. Healthy Living with Thalassemia. Available at: https://www.cdc.gov/ncbddd/thalassemia/living.html [accessed January 2023].
27. NHS. Thalassaemia – Living with. Available at: https://www.nhs.uk/conditions/thalassaemia/living-with/ [accessed January 2023].
28. Shah FT, et al. *Br J Haematol*. 2022;196(2):336–350.
29. Taher AT, Saliba AN. *Hematology Am Soc Hematol Educ Program.* 2017;2017(1):265–271.
30. Cappellini MD, Cohen A, Eleftheriou A, et al. Guidelines for the Clinical Management of Thalassaemia [Internet]. 2nd Revised edition. Nicosia (CY): Thalassaemia International Federation; 2008. Chapter 16, General Health Care and Lifestyle in Thalassaemia. Available from: https://www.ncbi.nlm.nih.gov/books/NBK173970/.
31. United Kingdom Thalassaemia Society. Available at: https://ukts.org/ [accessed January 2023].
32. Greek Thalassaemia Association. Available at: https://estha.gr/greek-thalassaemia-association/ [accessed January 2023].

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