What is Thalassemia Trait?

Cooley's Anemia Foundation
Leading the Fight Against Thalassemia
Introduction
引言
引言

This book contains basic information about the thalassemia trait.

Whether you have been diagnosed with the thalassemia trait or are simply interested in finding out more about it, we encourage you to consult your physician or a hematologist. If you carry the thalassemia trait and are considering having children, we also suggest that you seek the advice of a genetic counselor.

The Cooley’s Anemia Foundation’s message concerning the thalassemia trait is simple:

Being tested for the thalassemia trait is easy. And it is in everyone’s best interest, working in partnership with the health care professional of their choice, to be informed and take responsibility for their own health care.
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What is the thalassemia trait?

If you have been diagnosed with the thalassemia trait, this is simply another way of saying that you carry the genetic trait for thalassemia. A genetic trait is a kind of message or code contained in your body. You may pass this code on to your children, and they may pass it on to their children.

You may, for instance, carry the genetic trait for blue eyes, even if you yourself have brown eyes. Thalassemia trait is not a disease or condition but a kind of genetic possibility that you pass on to your descendants.

The thalassemia trait is not a disease.

Medical personnel often tell people who carry the thalassemia trait that they "have thalassemia" which can lead trait carriers to believe that they have some kind of health-threatening medical condition. This is not true. Medical terminology refers to the thalassemia trait as "thalassemia minor." So while it may be technically correct to say that trait carriers "have thalassemia" (or, more properly, "thalassemia minor"), trait carriers should be aware that carrying the genetic trait for thalassemia is not the same thing as having a disease.

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The thalassemia trait requires no medical treatment.

Physicians sometimes mistakenly prescribe iron supplements for thalassemia trait carriers, usually because they mistake the small size of the trait carrier's red blood cells with iron-deficiency anemia.

The only way to properly determine the need for iron supplements is to have a physician test iron levels in the patient's blood. Without a test of blood iron levels, iron supplements should not be prescribed for thalassemia trait carriers.

Why should I be concerned about the thalassemia trait?

Even though the thalassemia trait has no symptoms and cannot directly affect your health, it can indirectly affect your health and directly affect the health of your children. Doctors may mistake your thalassemia trait for a different condition and prescribe unnecessary and potentially harmful tests or treatments. Also, when two thalassemia trait carriers have a child, there is a one-in-four chance with each pregnancy that the child will be born with a serious blood disorder.

The thalassemia trait cannot become worse or turn into a serious disease.

Because the thalassemia trait is a genetic trait and not a health condition, it cannot "become worse" or change into one of the more serious forms of thalassemia that may require medical treatment.

Similarly, except for extremely rare cases, the thalassemia trait has no symptoms and requires no treatment.

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What is Thalassemia?

Thalassemia is the name of a group of genetic blood disorders. To understand how thalassemia affects the human body, you must first understand a little about how blood is made.

Blood carries oxygen from your lungs to other parts of your body. Oxygen is carried inside the red blood cells by a substance called hemoglobin. Hemoglobin is made of two different kinds of proteins, called alpha and beta globins. These globin proteins are made by genes located on pairs of chromosomes. If one or more of these globin-producing genes is abnormal or missing, there is a drop in globin production.
When only one beta globin gene or one or two alpha globin genes are missing (as is the case with trait carriers), the drop in globin production is so small it rarely affects the person's health.

But when two beta globin genes or three alpha globin genes are missing (as is the case with the more serious thalassemia disorders), the drop in globin production is so great that the red blood cells do not form properly and the blood cannot carry enough oxygen.

The result is anemia that begins in early childhood and lasts throughout life. When all four alpha genes are missing, the result is usually death in the womb.

Since thalassemia is not a single disorder but a group of related disorders that affect the human body in similar ways, it is important to understand the differences between the various types of thalassemia.
People who do not produce enough alpha globin protein have alpha thalassemia. It is commonly found in Africa, the Middle East, India, Southeast Asia, southern China, and occasionally the Mediterranean region.

Alpha globin is made by four genes, two on each strand of the chromosome 16. Individuals who have one or two abnormal alpha globin genes have alpha thalassemia trait.

An individual with alpha thalassemia trait defined by the presence of one abnormal alpha globin gene is said to have the silent carrier state. This condition, in which one of the four alpha globin genes is missing or defective, generally causes no health problems because the lack of alpha globin protein is so small that there is no anemia.

It is called "silent carrier" because of how difficult it is to detect. Silent carrier state is "diagnosed" by deduction when an apparently normal individual has a child with hemoglobin H disease or alpha thalassemia minor. It can also be diagnosed by special DNA testing.
If both parents carry alpha thalassemia minor in which the two abnormal genes of each parent are on opposite chromosomes, their children will inherit alpha thalassemia minor. 

An individual with alpha thalassemia trait defined by the presence of two abnormal alpha globin genes is said to have alpha thalassemia minor. 

Physicians often mistake alpha thalassemia minor for iron deficiency anemia and prescribe iron supplements that have no effect on the anemia. Both abnormal alpha globin genes may be on the same chromosome (cis position) or one may be on each chromosome in the pair (trans position).
The alpha thalassemia traits combine in different ways to produce blood disorders that range from mild to severe in their effect on the human body.

α-地中海貧血特質有不同的基因組合方式，因此所引起的血液疾病對人體的影響有輕有重。

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**if...**

*one parent has alpha thalassemia minor* with two abnormal alpha globin genes on the same chromosome

*雙親中一方為輕型α-地中海貧血，其兩個異常α珠蛋白基因位於同一染色體*

*and the other parent has the silent carrier state with a single abnormal alpha globin gene on one chromosome*

*雙親中另一方為無症狀特質，在一條染色體上有一個α珠蛋白基因異常*

*and the other parent has the silent carrier state with a single abnormal alpha globin gene on one chromosome*

*雙親中另一方為無症狀特質，在一條染色體上有一個α珠蛋白基因異常*

*there is a 25% chance with each pregnancy that their child will be born with hemoglobin H disease in which three of the four alpha globin genes are abnormal.*

*在這種情況下，α珠蛋白的缺乏足以引起中等至嚴重程度的貧血，甚至可能引起嚴重危害健康的疾病，如腎腫大，骨畸形和疲勞等。

**then**

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**normal hemoglobin**

25% 正常血紅蛋白

25% 正常血紅蛋白

**alpha thal trait**

25% α-地中海貧血特質

25% α-地中海貧血特質

**alpha thal silent carrier**

25% 無症狀 α-地中海貧血特質

25% 無症狀 α-地中海貧血特質

25% α-地中海貧血特質

25% α-地中海貧血特質
if...
如果...
both parents carry alpha thalassemia minor with two abnormal alpha globin genes on the same chromosome
父母均携带α地中海贫血轻微型，且至少有两条α珠蛋白基因异常，且位于同一条染色体上。

there is a 25% chance with each pregnancy that their child will inherit hydrops fetalis or alpha thalassemia major, in which all four alpha globin genes are abnormal.
每次妊娠，其后代均有可能携带α地中海贫血，且所有α珠蛋白基因均异常。有25%的可能性。此情况下，其后代将携带严重的α地中海贫血或胎水肿。

most individuals with this condition die before or shortly after birth. In some extremely rare cases, in utero blood transfusions have allowed the birth of children with hydrops fetalis who then require lifelong blood transfusions and medical care.
大多数携带这种疾病的个体在出生前或出生后不久即死亡。在极少数的极其罕见情况下，宫内输血可能使一部分胎水肿的个体出生，之后还需终生接受输血和医疗护理。

...then
...那麼

...那様

25% hydrops fetalis
胎兒水肿

25% alpha thal major
α地中海贫血

25% normal hemoglobin
25% 正常血紅蛋白

25% alpha thal trait (cis Type)
50% α地中海貧血特質（順式）
Beta Thalassemia

People who do not produce enough beta protein have beta thalassemia. It is found in people of Mediterranean descent such as Italians and Greeks, and is also found in the Arabian Peninsula, Iran, Africa, Southeast Asia, and southern China. Beta globin is made by two genes, one on each chromosome 11. Individuals who have one abnormal beta globin gene have beta thalassemia trait.

BETA THALASSEMIA MINOR or BETA THALASSEMIA TRAIT.
In beta thalassemia trait, one of the two genes is abnormal but the lack of beta protein is not great enough to cause problems in the normal functioning of the hemoglobin. A person with this condition simply carries the genetic trait for beta thalassemia and will usually experience no health problems other than a mild anemia. As in alpha thalassemia minor, physicians often mistake the small red blood cells of the person with beta thalassemia minor as a sign of iron-deficiency anemia and incorrectly prescribe iron supplements.

β-地中海贫血

在β-地中海贫血中，两个基因中的一个异常，但是β蛋白缺乏不严重，不足以影响血红蛋白的正常功能。
both parents carry the beta thalassemia trait.

父母双方都携带β-地中海贫血特质

There is a 25% chance with each pregnancy that their child will inherit two abnormal beta globin genes. In its most severe form, this may cause beta thalassemia major or Cooley’s anemia, a blood disorder in which the lack of beta protein causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care. Lifelong transfusions lead to iron overload which must be treated with chelation therapy to prevent early death from organ failure.

In a somewhat milder form, the inheritance of two abnormal beta globin genes may cause beta thalassemia intermedia, in which the lack of beta protein in the hemoglobin causes a moderately severe anemia and significant health problems including bone deformities and enlargement of the spleen.

Due to the wide range in severity of this condition, the borderline between thalassemia intermedia and thalassemia major can be confusing. When a patient is more dependent on blood transfusions he is likely to be classified as thalassemia major.

每对父母各携带β-地中海贫血特质。在每个怀孕中，有25%的机会其孩子会继承两个异常的β-珠蛋白基因。在最严重的形式，这可能导致β大地中海贫血或Cooley's贫血，是一种血红蛋白缺乏的贫血症，需要定期输血和持续的医疗护理。长期输血会引发铁质过量，必须通过螯合药来预防早期器官衰竭而早死。

在稍轻的形态，两个异常的β-珠蛋白基因的存在会引起β中轻型地中海贫血，血红蛋白缺乏将引发中等程度的贫血，并导致一些显著的健康问题，包括骨骼变形和脾肿大等。

由于这个条件的严重范围很大，β中间型和重型地中海贫血之间的界限模糊。如果一个孩子对输血的依赖性更弱，那么他更可能是患有中轻型地中海贫血。

there is a 25% chance with each pregnancy that their child will inherit two abnormal beta globin genes. In its most severe form, this may cause beta thalassemia major or Cooley’s anemia, a blood disorder in which the lack of beta protein causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care. Lifelong transfusions lead to iron overload which must be treated with chelation therapy to prevent early death from organ failure.

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如果...
The beta thalassemia trait can also combine with "variant" hemoglobins to produce other related blood disorders.

β-地中海贫血特質也可能與「變異」血紅蛋白組合，引起其他相關的血液疾病。

β-地中海贫血特質也可能與“變異”血紅蛋白組合，引起其他相關的血液疾病。

If...
如果...

one parent carries the beta thalassemia trait
如果雙親中一方為輕型β-地中海貧血特質者
and the other parent carries the hemoglobin E trait
另一方為血紅蛋白E特質者

25% normal hemoglobin
25% 正常血紅蛋白
25% 輕型β-地中海貧血
25% E beta thalassemia
25% 血紅蛋白E beta thalassemia

and the other parent carries the hemoglobin E trait
另一方為血紅蛋白E特質者

25% E beta thalassemia
25% 血紅蛋白E beta thalassemia
25% 血紅蛋白E特質
25% β-地中海貧血特質

25% 血紅蛋白E特質
25% β-地中海貧血特質

there is a 25% chance with each pregnancy that their child will be born with E beta thalassemia, a moderately severe anemia that has similar symptoms to beta thalassemia intermedia but on occasion may be as severe as thalassemia major.

Hemoglobin E is one of the most common abnormal hemoglobins. It is usually found in people of Southeast Asian ancestry, such as Cambodians, Vietnamese and Thai.

...then
...那麼
one parent carries the beta thalassemia trait
另一方為β-地中海貧血特質者
double one parent carries
the hemoglobin S trait (the abnormal hemoglobin found in people with sickle cell disease)
另一方為血紅蛋白S（在镰状細胞病患者中發現的一種異常的血紅蛋白）特質者
and the other parent carries the beta thal trait
另一方為血紅蛋白S（在镰状細胞病患者中發現的一種異常的血紅蛋白）特質者

there is a 25% chance with each pregnancy that their child will be born with sickle beta thalassemia. Hemoglobin S is commonly found in people of African or Mediterranean ancestry, such as Africans, Italians, Greeks, Turks, and in people from the Caribbean.
The severity of the condition varies according to the amount of normal beta globin produced by the beta gene. When no beta globin is produced by the beta gene, the condition is almost identical to sickle cell disease. When some beta globin is produced by the beta gene, the condition is less severe.

...那麼
每次妊娠，他們的後代將有25%的機率患有遺傳性镰状細胞——β-地中海貧血。血紅蛋白S經常在非洲和地中海後裔中發現，如非洲人、意大利人、希臘人、土耳其人和加勒比海人。

25% sickle-beta thalassemia
25% 鰭狀細胞
β-地中海貧血
25% 鰭狀細胞
β-地中海貧血
25% 正常血紅蛋白
正常血紅蛋白
25%
25% 正常血紅蛋白
正常血紅蛋白
25%
25% 正常血紅蛋白
正常血紅蛋白
25%
Who Carries the Thalassemia Trait?
哪些人具有地中海贫血特質？
哪些人具有地中海贫血特質？

It is estimated that over 2 million people in the United States carry the genetic trait for thalassemia.

在美國，估計有超過兩百萬名地中海貧血基因攜帶者

Thalassemia was originally believed to be common only to people of the Mediterranean region, such as Italians, Greeks and Turks. (An early name for thalassemia major or Cooley’s anemia was Mediterranean anemia.)

地中海貧血最初被認為是一種只發生於地中海地區（如義大利、希臘和土耳其等地）的一種疾病，故重型地中海貧血或庫利貧血症在早期被稱為地中海熱。

地中海貧血最初被认为是一种只发生于地中海地区（如意大利、希腊和土耳其等地）的一种疾病，故重型地中海贫血或库利贫血症在早期被称为地中海热。
Since then, scientists have discovered that the thalassemia trait is found in people of many other regions, including the Arabian Peninsula, Africa, the Indian subcontinent, China, Southeast Asia, and the Caribbean.

Today, due to the migration and intermarriage of different ethnic populations, the trait for thalassemia is found in people with no obvious ethnic connection to the disorder. For this reason, the New York State Department of Health provides couples seeking a marriage license with information about thalassemia.

後來，科學家發現地中海貧血特質也發生於其他地方，如阿拉伯半島、非洲、印度次大陸、中國、東南亞和加勒比海地區等。

今天，由於不同種族人口的遷移和通婚，地中海貧血特質已遍布於全世界，甚至在這種疾病並沒有明顯種族聯繫的人群中也有發現。基於這種原因，紐約州衛生部向想要獲取結婚證的未婚夫婦提供有關地中海貧血的資訊。

后来，科学家发现地中海贫血特质也发生于其它地方，如阿拉伯半岛、非洲、印度次大陆、中国、东南亚和加勒比海地区等。

今天，由于不同种族人口的迁移和通婚，地中海贫血特质已遍布于全世界，甚至在与这种疾病并没有明显种族联系的人群中也有发现。基于这种原因，纽约州卫生部向想要获取结婚证的未婚夫妇提供有关地中海贫血的信息。
Finding out if you have the genetic trait for thalassemia begins by determining the size of your red blood cells.

If you have a routine blood test known as a Complete Blood Count, or CBC, already on file at your doctor’s office, ask your doctor to look at the Mean Corpuscular Volume, or MCV. The MCV reading determines the size of your red blood cells. For adults, if the MCV reading is less than 75 and you are not iron deficient, you may be a trait carrier. For children, the MCV reading may be lower and varies according to their age.

If your MCV reading indicates that you may have the thalassemia trait, your doctor should then perform additional tests to confirm that you have the thalassemia trait and to determine what kind.

Although the MCV reading is a good indicator of whether a person may have either the alpha or the beta thalassemia trait, finding out for certain if you have either trait involves additional tests.
Special tests called hemoglobin electrophoresis and quantitation of hemoglobin A2 and hemoglobin F are a reliable way of determining whether or not a person has the trait for beta thalassemia (beta thalassemia minor). These tests are available at most large hospitals and clinics.

Testing for alpha thalassemia trait (alpha thalassemia minor) is usually done by a process of exclusion; people who have low MCV (not due to iron deficiency), a normal hemoglobin electrophoresis, quantitative hemoglobin A2 and quantitative hemoglobin F, and are of the appropriate ethnic origin are presumed to have alpha thalassemia minor.

In some circumstances, more definitive "molecular" testing is performed to determine the presence or absence of thalassemia trait. As these genetic tests are developed further, they will be used more widely to test for thalassemia trait.

Remember that the first step to finding out if you have the thalassemia trait is easy. Just call your doctor and ask him or her to check your MCV reading. Testing for thalassemia can be done at most hospitals. When you go in for your test bring this book with you to share with your doctor.
If you have alpha or beta thalassemia trait and are considering having a child or are already pregnant, your partner should be tested to see if he or she has the thalassemia trait. If you both have thalassemia trait, there are several things you can do.

Inform your obstetrician about your thalassemia trait. Discuss what it might mean for your unborn child. If necessary, share this book with your doctor.

If you want to determine whether your unborn child has any form of thalassemia, there are two kinds of tests you can request.
AMNIOCENTESIS

Amniocentesis is performed in the second trimester of pregnancy, after about 15 weeks of gestation. Using ultrasound as a guide, the doctor withdraws 2-3 tablespoons of amniotic fluid from the mother’s womb through a very thin needle inserted in the mother’s abdomen. Fetal cells that are floating free in the amniotic fluid are then analyzed for the thalassemia mutations.

CHORIONIC VILLUS SAMPLING (CVS)

CVS can be performed somewhat earlier than amniocentesis, at about 10-11 weeks of pregnancy. In this test, the doctor removes a small sample of the chorionic villi, or the cells that will form the placenta. The cells are removed either with a thin needle inserted in the mother’s abdomen or with a thin catheter inserted in the vagina. These cells, which contain the same genetic information as the fetus, are analyzed for the thalassemia mutations.

If you are interested in either of these tests, ask your obstetrician to refer you to a prenatal testing center.

羊膜穿刺

羊膜穿刺术在妊娠期的第二个三个月期间中，也就是怀孕第15周左右进行。在超声引导下，医生将一个非常细的针头插入孕妇腹部，从孕妇的子宫中取出2-3大汤匙的羊水。然后检查漂浮在羊水中的胎兽数细胞，以确定胎儿是否有地中海贫血突变。

绒膜绒毛取样（CVS）

绒膜绒毛取样的时间可比羊膜穿刺术早，大约在怀孕第10-11周就可以进行。医生将一个细针头插入孕妇腹部，或将一根细导管插入孕妇阴部，取出少量的绒膜绒毛或将要形成胎盘的细胞，然后用这些携带与胎儿相同遗传信息的细胞进行地中海贫血突变分析。

如果您对这两种检查中的任何一种感兴趣，请让您的产科医生为您推荐一间产前检查中心。

绒膜绒毛取样（CVS）

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About the Cooley’s Anemia Foundation

The Foundation’s mission is:

- Advancing the treatment and cure for this genetic blood disease.
- Enhancing the quality of life of patients.
- Educating the medical profession, thalassemia trait carriers and the public about thalassemia.

基金會的使命是:

- 促进遗传性血液疾病的治疗。
- 提高患者的生活素质。
- 為醫療界人士、地中海貧血特質者和公眾提供關於地中海貧血病的教育。

For more information about thalassemia and what the Cooley’s Anemia Foundation is doing to fight it, call 800-522-7222 or visit our website (www.cooleyasanemia.org).

如果您想了解更多關於地中海貧血的資訊，或瞭解庫利貧血基金會如何抗擊地中海貧血，請撥800-522-7222，或訪問我們的網站（www.cooleyasanemia.org）。

如果您想了解更多关于地中海贫血的信息，或了解库利贫血基金会如何抗击地中海贫血，请拨800-522-7222，或访问我们的网站（www.cooleyasanemia.org）。
This publication was made possible by grants from the New York State Department of Health and the Centers for Disease Control and Prevention.

Written by David Surface.

Designed by MediumBlend.

Our thanks to Dr. Alan Cohen of Children’s Hospital of Philadelphia for his invaluable assistance in preparing this publication.

感謝費城兒童醫院 Alan Cohen 博士對本書出版的大力支持

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