HOW CAN YOU KNOW IF YOU ARE A THALASSAEMIA CARRIER?

Assessing your thalassaemia status is easy!

1. You can start with basic laboratory tests like a complete blood count (CBC), which measures the quantity and size of red blood cells.
2. Following that, more specific tests are necessary, such as liquid chromatography (HPLC), a precise and reliable method for identifying the thalassaemia trait, haemoglobin electrophoresis, which assesses haemoglobin levels and identifies abnormal types of haemoglobin, or genetic tests that will analyse your DNA. All of these can be done with a simple blood test!
3. A trained healthcare professional will evaluate the results and provide appropriate guidance on your condition and available options.

THE FUTURE OF DIAGNOSIS AND SCREENING

Significant advances in thalassaemia screening and diagnostics have enhanced accuracy, efficiency, and accessibility. Molecular genetic testing has notably improved with Next-Generation Sequencing (NGS), accelerating the identification of mutations. Additionally, Point-of-Care Testing (POCT) devices have emerged as a critical resource, especially in resource-limited regions, by quickly identifying at-risk individuals for thalassaemia.

These technologies are not only instrumental in carrier detection but also in the diagnosis of affected individuals, including prenatal diagnosis. Non-Invasive Prenatal Testing (NIPT), which analyses cell-free foetal DNA in maternal blood, has been introduced. However, its practical application remains limited.

GETTING TESTED IS VITAL FOR:

- **YOUR HEALTH:** Being a thalassaemia carrier means you will, most likely, not experience any health problems, except perhaps a mild anaemia. This condition can often be mistaken for iron-deficiency anaemia. Accurate knowledge of your carrier status is crucial to avoid misdiagnosis or inappropriate treatment.
- **YOUR FAMILY:** If you are a thalassaemia carrier, this means that thalassaemia runs in your family. Therefore, it is advisable for your parents and other relatives to consider testing as well.
- **YOUR CHILDREN:** Should both you and your partner be carriers of thalassaemia, there is a 25% risk with each pregnancy that your child that your child will be born with thalassaemia major, the most severe form of the condition.

For more information, you can visit
www.thalassaemia.org.cy

THALASSAEMIA INTERNATIONAL FEDERATION

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THALIA

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THALASSAEMIA CARRIER CHECK: Get Informed, Get Tested

THALASSAEMIA TYPES AND THEIR SIGNIFICANCE TO HEALTH

Thalassaemia is a group of inherited blood disorders characterised by decreased production of haemoglobin, the protein that carries oxygen in red blood cells.

There are two different types of thalassaemia; alpha (α-) and beta (β-) thalassaemia. Thalassaemia is commonly classified as either thalassaemia minor, thalassaemia intermedia, and thalassaemia major to indicate the severity of the condition.

Individuals with thalassaemia minor, sometimes known as 'carriers', are generally healthy. Although they may have a mild form of anaemia, they typically do not exhibit any symptoms and, as such, do not undergo medical treatment.

Thalassaemia intermedia or non-transfusion-dependent thalassaemia is of intermediate gravity between thalassaemia minor and β-thalassaemia major. Due to its clinical diversity, thalassaemia intermedia patients may occasionally need blood transfusions.

In contrast, individuals with β-thalassaemia major or transfusion-dependent thalassaemia, the most serious form of the disease, suffer from severe, life-threatening anaemia, usually leading to poor growth, substantial health complications and a shortened lifespan, if left untreated.

GLOBAL PREVALENCE OF THALASSAEMIA CARRIERS

Have you heard of thalassaemia? If not, then you might be surprised to know that it is much more common than you think.

7% of the global population are estimated to be carriers of haemoglobin disorders, including thalassaemia. In several areas of the world, the thalassaemia carriers' prevalence can even exceed 20%.

Carriers are predominantly found in countries surrounding the Mediterranean Sea—such as Cyprus, Greece, Italy, Lebanon, Syria, and Egypt—as well as other North African nations. The Middle East also has a significant number of carriers, particularly on the Arabian Peninsula and in countries like Iran and Iraq. Furthermore, the condition is prevalent in the Indian subcontinent and Southeast Asia, including Thailand, Indonesia, Vietnam, and the Maldives, as well as the Western Pacific regions of China, Malaysia, and the Philippines.

Being a Thalassaemia carrier does not mean that you have the disease.

It means you carry one of the faulty genes that cause the disorder, but you will not develop thalassaemia at any point.

WHY IS IT IMPORTANT TO KNOW IF YOU ARE A CARRIER?

Thalassaemia is an inherited condition; this means that it is passed down from parents, both of whom are carriers, to their children. Carriers typically do not require any treatment or intervention during their lifetime.

In fact, thalassaemia carriers are often unaware of their status and would not suspect it without specific blood testing.

When both parents carry the β-thalassaemia gene, each of their pregnancies has a 25% chance of resulting in a child with β-thalassaemia major—the most serious form of the disease. Such children must receive lifelong medical care that includes regular blood transfusions, iron chelation therapy to remove excess iron, and comprehensive multidisciplinary care.

INHERITANCE OF THALASSAEMIA