GENE SOLUTIONS

#BeThalassemiAware
with Gene Solutions
2024

- What is Thalassemia?
- Why should we care?
- How do we know we're carrying thalassemia traits?
- How do we prevent?
- What's next after the result?

#beThalassemiAware

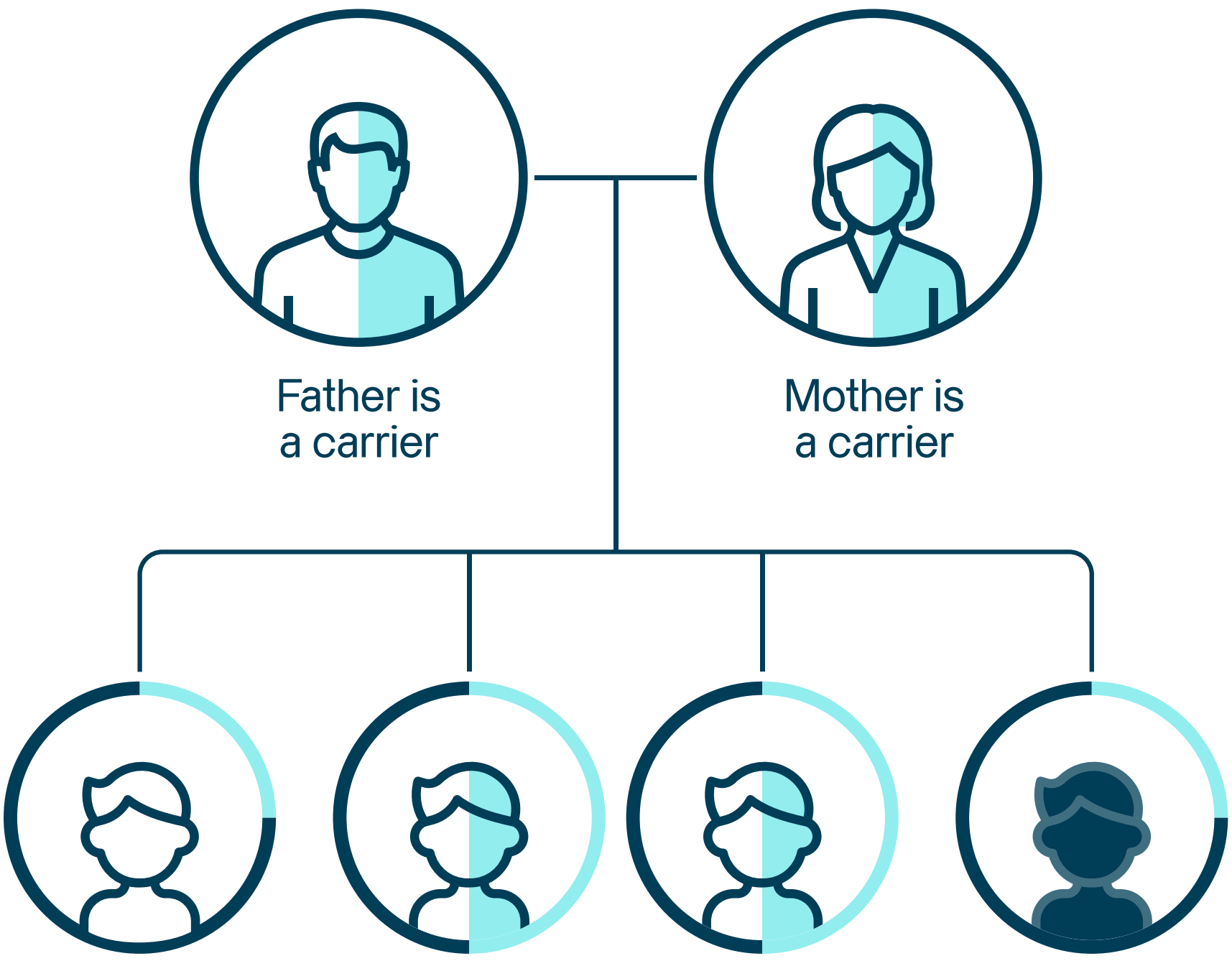
to empower
you and
your family

What is Thalassemia?

- Thalassemia, an inherited hemoglobinopathy, is one of the most common genetic disorders in the world that can pass from parents to children.
- There are 2 main types of thalassemia; alpha (α) and beta (β) –thalassemia – but when both parents have the same thalassemia mutation genes, here's a chance that their child will have more severe symptoms.

25% Risk of thalassemia major when both parents carry the same (α or β) –thalassemia mutation genes.

Recessive Inheritance



25% chance the child will be unaffected (healthy)

50% chance the child will be a carrier (minor)

25% chance the child will be severely sick (major)

Due to the risk of passing genetic disorders such as Thalassemia, pregnant women are advised to consult with a doctor.

Thalassemia **major** and **minor**

Thalassemia **major** and **minor** manifest varying degrees of symptom severity, ranging from potentially life–threatening to manageable with early identification and intervention

MINOR

Subclinical

Absence of any symptoms, absence of anemia (suitable for blood donation).

Mild

Symptoms of anemia are often subtle.

Moderate

Anemic symptoms become noticeable in children older than 6 years old.

MAJOR

Severe

Anemic symptoms manifest in children under 2 years old; enlarged liver and spleen, yellowing of the skin, delayed physical and motor development. Facial features may be affected: flat nose, prominent forehead and cheekbones, protruding jawbone.

Very Severe

Fetal hydrops may occur during pregnancy, leading to immediate postnatal death due to heart failure and severe anemia.

The main treatment for Thalassemia major is blood transfusions.

If thalassemia trait runs in your family, consult a doctor if you begin planning a family. Thalassemia minor doesn't typically cause serious symptoms so you may not realize you're a carrier.

Guidelines for the management of thalassemia (2023) – THALASSEMIA INTERNATIONAL FEDERATION

Why should we care?

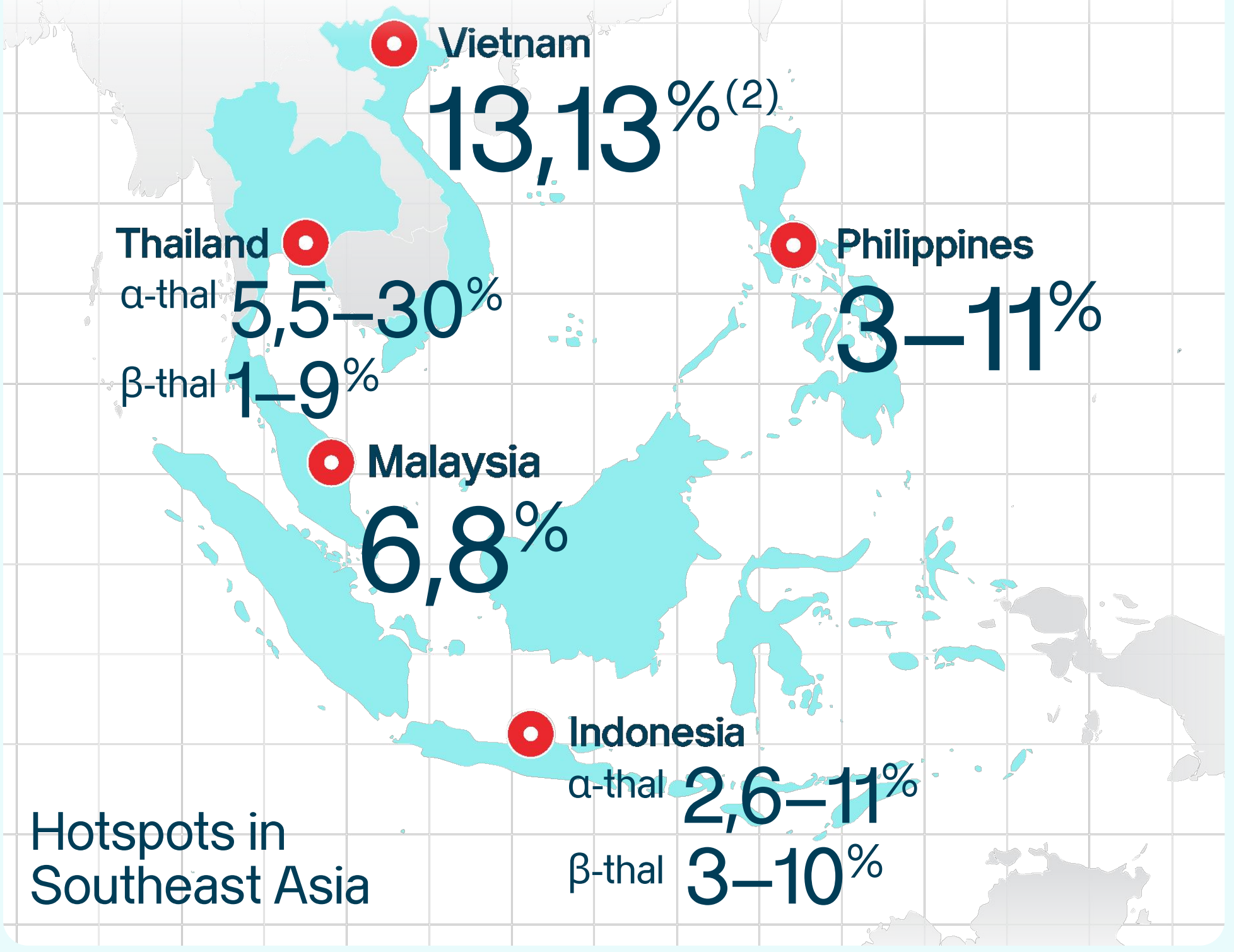
Understanding Thalassemia is crucial for effective management and support for you and your loved ones.

Approximately **5–7% of the world population** carries the Thalassemia gene.(1)

Individuals of Asian descent are at **heightened risk.**

56,000 newborns are affected by Thalassemia major each year.(1)

Southeast Asia belongs to the **Thalassemia Belt** due to a high prevalence of β-thalassemia



(1) Global Thalassemia Review 2023 – THALASSEMIA INTERNATIONAL FEDERATION

(2)
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How do we know we're carrying thalassemia traits?

01

Blood & carrier screening test

We can take a carrier screening test for Thalassemia at any stage of life. However, due to a lack of awareness, many people don't undergo screening unless prompted by considerations such as pregnancy or marriage.

02

ACOG/SMFM recommends NIPT as part of all pregnancy screen*

- Non–invasive Prenatal Testing (NIPT) has emerged to aid in thalassemia carrier screening for the mother.
- **Relevant**: NIPT not only informs the baby genetic status but also detects the thalassemia gene of the mother. This enables doctors to provide genetic counseling as early as 9 weeks into the pregnancy.
- **Improved Accuracy**: Utilizing Next–Generation Sequencing technology, the NIPT test surpasses traditional blood tests in detecting relevant genes with heightened accuracy and sensitivity.

*ACOG/SMFM clinical management guidelines for obstetricians and gynecologists released Practice Bulletin 226, "Screening for Fetal Chromosomal Abnormalities," in October 2020 which advocates all pregnancies be screened with NIPT. This guideline endorses NIPT as the most sensitive and specific prenatal screening option versus traditional screening methods.

How do we manage the risk?

01

Community health empowerment – spark awareness

Share the risks and prevention of Thalassemia in your community.

02

Empowering Carrier Screening

Identify carriers through genetic testing for informed family planning decisions.

03

Genetic Counseling/ Testing

Get expert guidance on premarital and prenatal decisions, offering support and information and available interventions.

04

Get NIPT integrated with thalassemia carrier screening at first trimester of pregnancy

Contact your local health providers for guidance.

What's next after the result?

Negative

Not a carrier of thalassemia mutation. Continue pregnancy with standard prenatal care.

Positive as a Carrier of Thalassemia

- Seek Genetic counseling/ testing for father
- If both parents carry the same thalassemia mutations, your doctor will guide you on pre/ post–natal diagnostic tests for the baby.
- Discuss with your healthcare provider for treatment and prenatal care options based on genetic results.

Pregnancies involving major thalassemia are often high risk with adverse health outcomes for both mother and fetus. Therefore, we should:

- Improve prognosis with NIPT integrated with thalassemia screening.
- Prompt diagnosis to enable earlier treatment at post–birth.

Additionally, the results gain insights into family planning for subsequent pregnancies.

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Finding out if you're a carrier of Thalassemia is important for planning your family and getting the right care.

Contact:
info@genesolutions.com
www.genesolutions.com

Simply reach out to your healthcare provider or Gene Solutions to learn more.

Scan QR code to take our knowledge Quiz and get updated information on NIPT – integrated with thalassemia carrier screening