Thalassemia HubKit

"At age nine, I chose to not let thalassemia defeat me and to live a happy life. I've always tried to take responsibility and respect to the next level in managing my health care." ~ Maria Hadjidemetriou, person living with thalassemia

Diagnostic Inequities

- An estimated 2,000,000 Americans carry the genetic trait for thalassemia.
- Thalassemia affects an estimated 5-7% of the worldwide population and is more common among people of Mediterranean, Middle Eastern, South Asian, Southeast Asian, Chinese, Northern African, and Caribbean backgrounds.
- 8 out of 10 patients with thalassemia live in low- and middle-income countries.
- In most countries with medium- and high-disease prevalence, patients with beta (β)-thalassemia major do not reach the age of 20 years.
- More than half a million children are expected to be born with thalassemia and other hemoglobin disorders worldwide by 2030.

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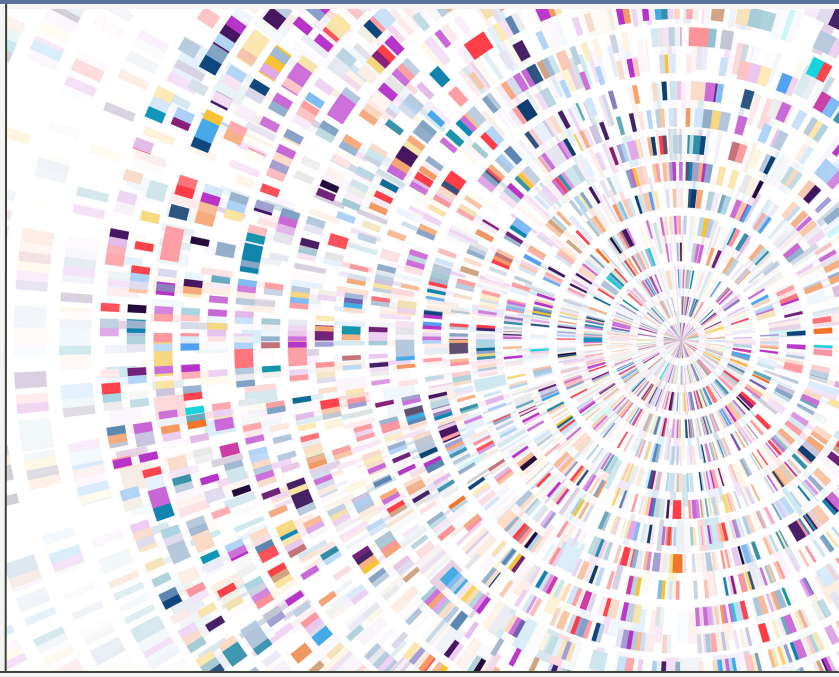

Tests and Diagnostic Tools

- **Carrier/Trait testing**: should be done if you and your partner both carry the thalassemia trait; there's a 1-in-4 chance that your baby will have a severe form of the disorder.
  - Involves a **Complete Blood Count (CBC)**, make sure your doctor looks at the **Mean Corpuscular Volume (MCV)** of your CBC. If the MCV reading is less than 75, and if you are NOT iron deficient, you may be a trait carrier.
  - Additional tests such as **hemoglobin electrophoresis** (which measures different types of hemoglobin in the blood) and **quantification of hemoglobin A2 and hemoglobin F** will help determine if you carry the trait.
  - You may see a genetic counselor based on the outcome of carrier/trait testing to help inform your reproductive choices, ensure appropriate maternal care and facilitate diagnosis in newborns.
- **Newborn screening**: the CDC recommends screening newborns for alpha (α) and beta (β) thalassemia; Primary screening is generally performed using structural hemoglobin testing like **high-performance liquid chromatography (HPLC)** or **isoelectric focusing (IEF).**
  - Results of the screening are confirmed through **genetic testing** using **Multiplex Ligation-Dependent Probe Amplification (MLPA)** and/or sequencing. Genetic testing can also be used to follow up on inconclusive results, identify mild mutations for carrier screening purposes, investigate genetic interactions, and for prenatal testing.

Source

Access Barriers

- Lack of healthcare professionals to accurately diagnose the condition early on.
- Patient location, particularly if they live in a low- to medium-income country.
- Patient access to coordinated treatment programs or hematologists.
- Financial costs to see hematologist or undergo genetic testing.
- Lack of knowledge about symptoms and/or screening options.

Call to Action

Read up. Keep reading to increase your awareness and understanding of diagnostic testing.

Learn. If you have a disease or health problem, learn about the specific tests and how they may impact your health and treatment outcomes.

Understand. Know what's normal and abnormal for you.

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