

# Thalassemia

Coleen Salazar  
Midwives College of Utah  
BIOL 2010: Genetics  
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# Objectives

01

**Define**

What? Who? Where?

02

**Genetics**

Variant?

03

**Screening**

Why? How?

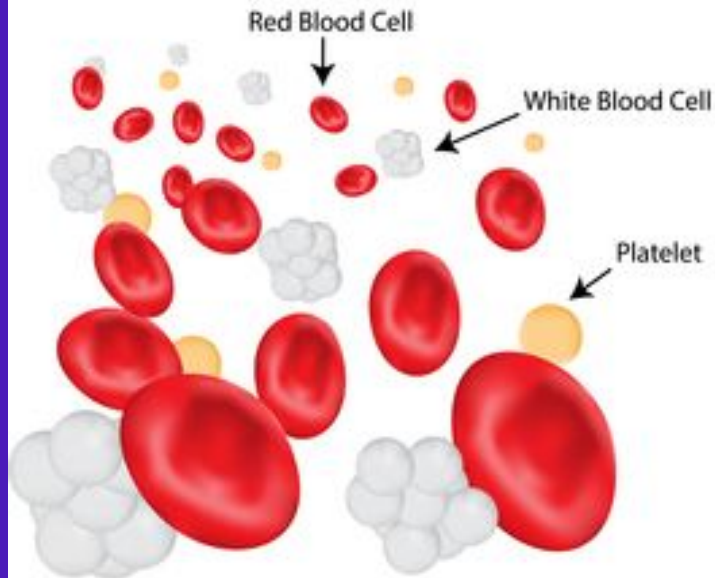
The background is a vibrant purple with several large, organic, wavy shapes in shades of pink, red, and blue. Three small white circles are scattered across the page: one in the upper left, one in the lower left, and one in the middle right.

**01**

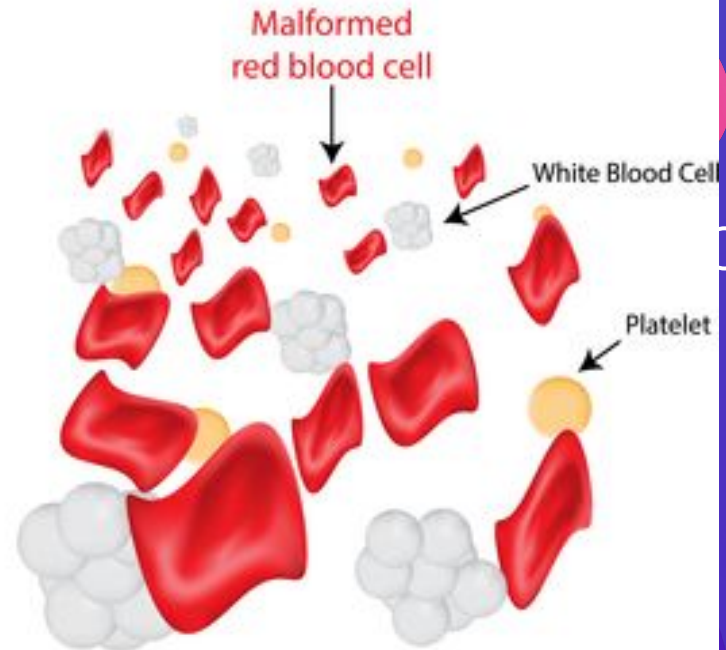
# Definition

# Thalassemia

## Normal



## Thalassemia





**100,000**

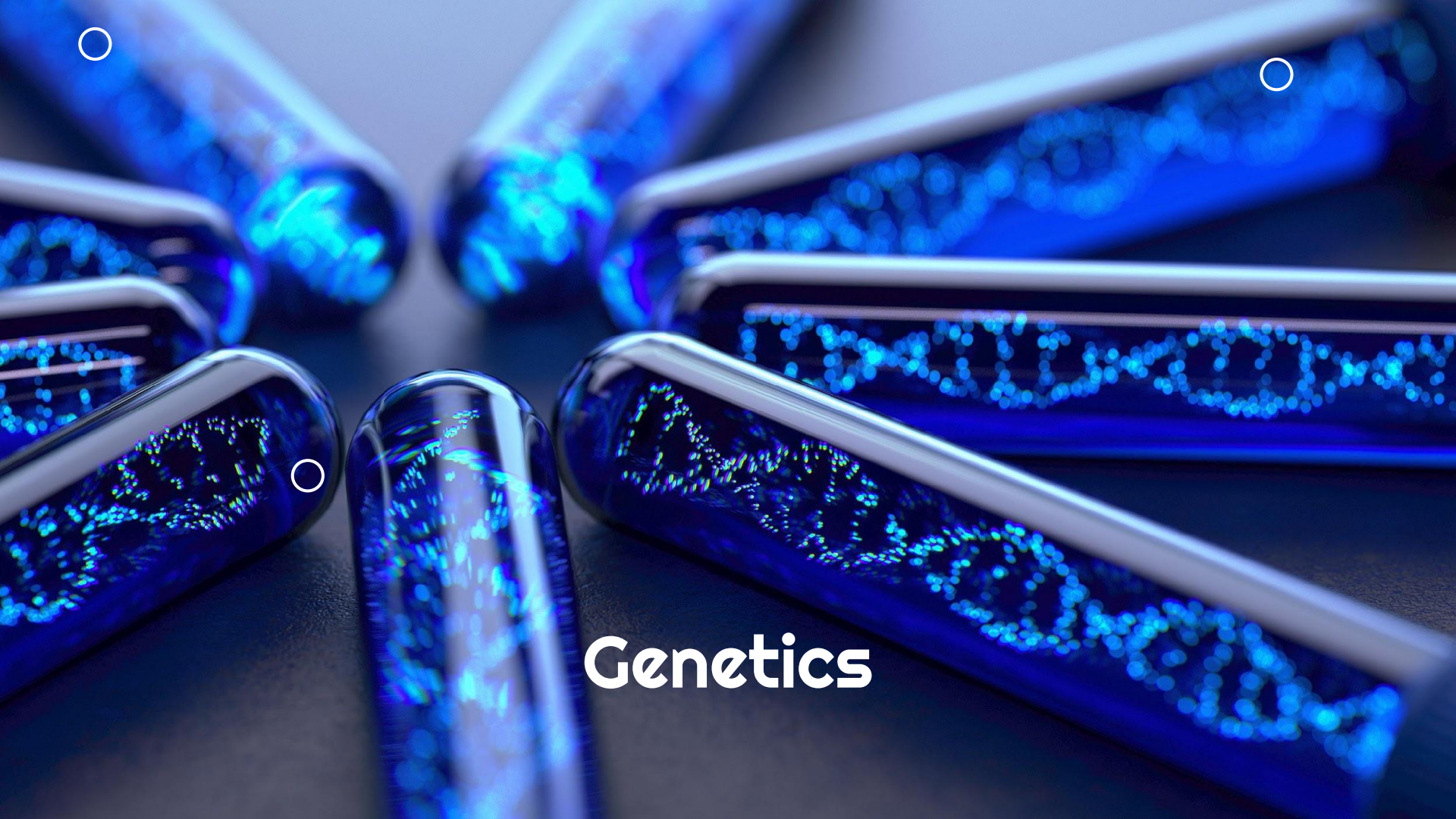
Babies worldwide

# Populations at Risk



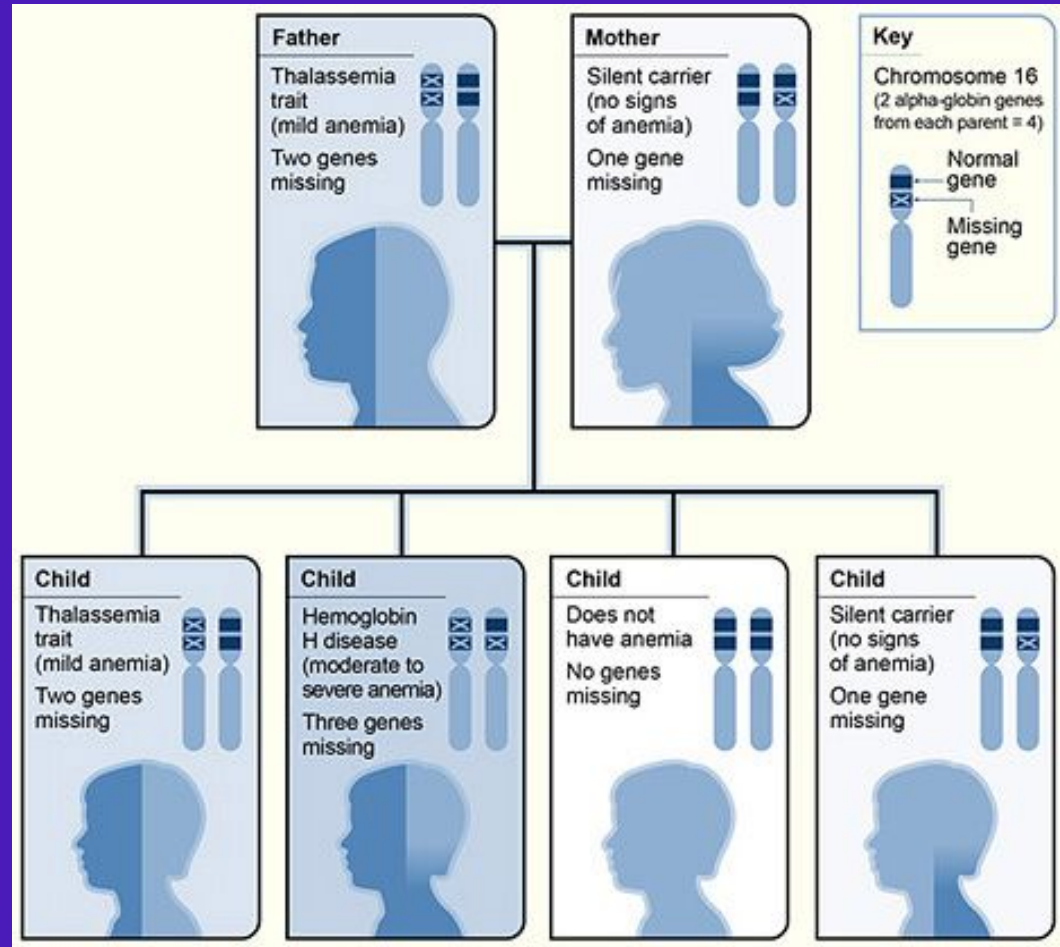
- South Asian
- Italian
- Greek
- Middle Eastern
- African





# Genetics

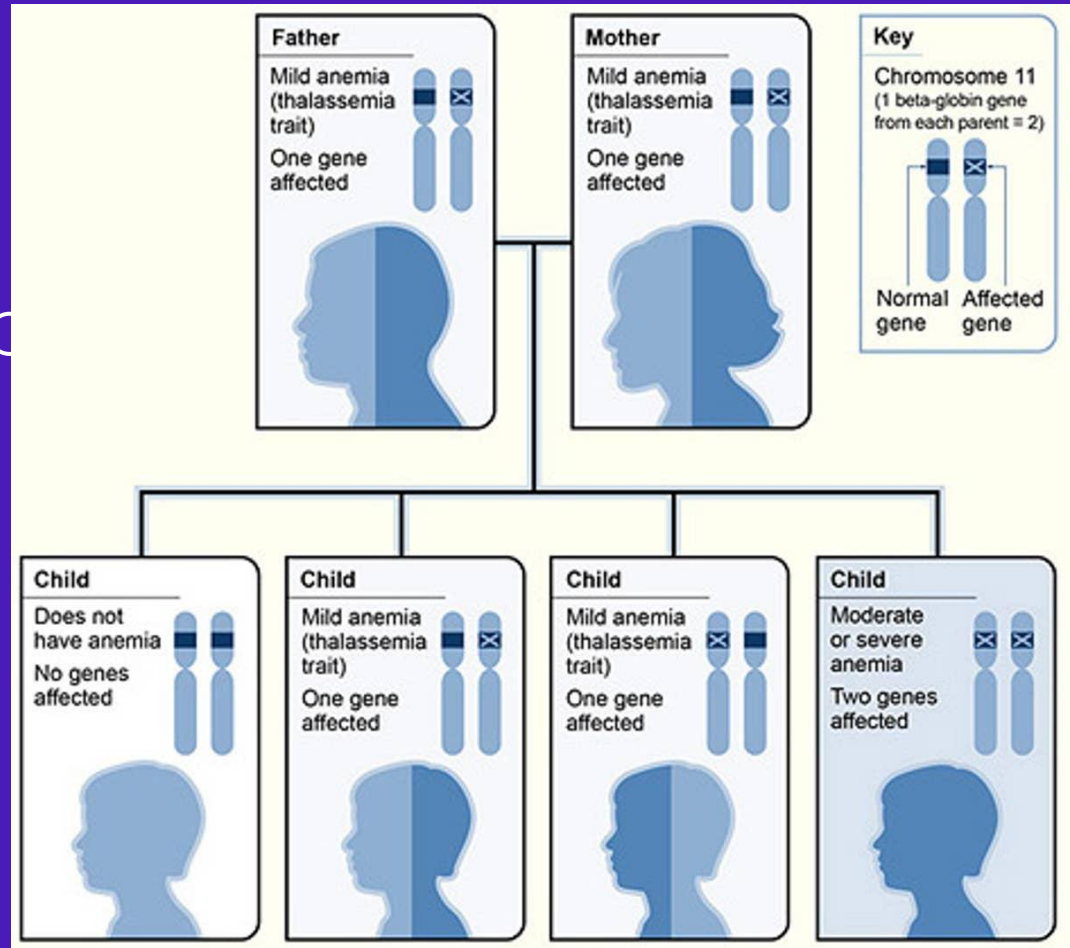
# Inheritance Pattern $\alpha$ - Thalassemia





# Inheritance Pattern

## $\beta$ - Thalassemia



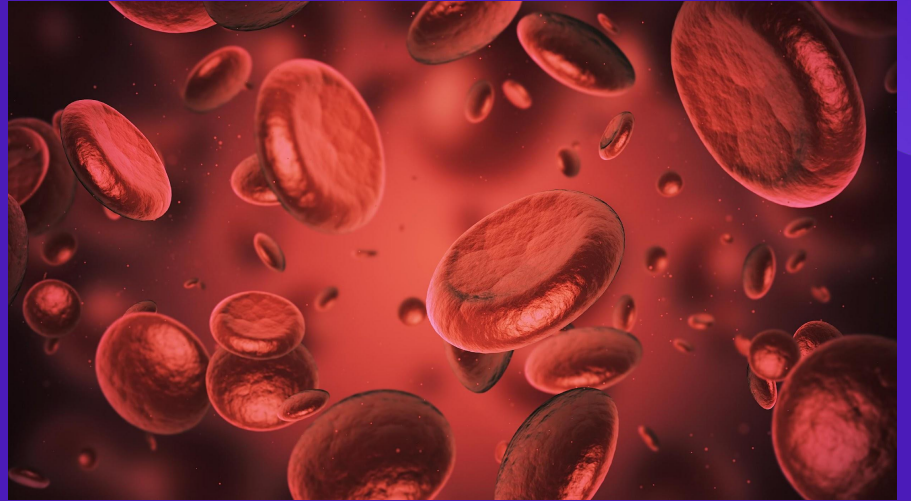
# Symptoms:

- Feeling tired or weak
- Shortness of breath
- Paleness
- Dizziness and fainting
- Headaches



# Treatment

- No tx
- Supplementation of folic acid
- Iron chelation therapy
- Red blood transfusions



03

# Screening

# Screening Considerations

## Complete Blood Count (CBC)

Anemia markers?

## HgB Electrophoresis

Different types of HgB

## Cell-free DNA (cfDNA)

Available anytime after 10th wk GA

## Pre-conception

Genetic screening or both partners/at risk



# Diagnostic

## Chorionic Villa Sampling

- 1st trimester
- Fetal Karyotype



# References

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Hudecova, I., & Chiu, R. W. (2017). Non-invasive prenatal diagnosis of thalassemias using maternal plasma cell free DNA. *Best practice & research. Clinical obstetrics & gynaecology*, 39, 63–73.  
<https://doi.org/10.1016/j.bpobgyn.2016.10.016>

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<https://www.nhlbi.nih.gov/health/thalassemia/pregnancy>

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<https://doi.org/10.1590/1519-6984.246062>

Chen, C., Li, R., Sun, J. et al. Noninvasive prenatal testing of  $\alpha$ -thalassemia and  $\beta$ -thalassemia through population-based parental haplotyping. *Genome Med* 13, 18 (2021).  
<https://doi.org/10.1186/s13073-021-00836-8>

U.S. Centers for Disease Control and Prevention - Division of Blood Disorders (2022, June 2) NB Screening for Thalassemia in the U.S. [Webinar] <https://youtu.be/KWjU30W7qSY>

# Thanks!

Do you have any questions?  
Coleen.Salazar@midwifery.edu  
760-644-1826

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