

WHAT IS BETA-THALASSEMIA?

Beta-thalassemia is a rare, inherited blood disorder caused by a genetic defect in hemoglobin, which resides in red blood cells (RBCs) and carries oxygen throughout the body. This defect leads to the production of fewer and less healthy RBCs and can lead to severe anemia and other serious symptoms.^{1,2}

Types^{2,3,4}

Beta-thalassemia is often divided into 2 categories



MAJOR

Symptoms appear within the first two years of life



INTERMEDIA

Typically presents in pre-adolescence

Additionally, some people may carry the gene for thalassemia, but be asymptomatic. This is known as trait or minor.

Symptoms^{2,3}

Major and Intermedia may present with common symptoms, such as:



Anemia



Bone and muscle abnormalities



Growth deficiencies



Abnormalities and complications of the spleen, liver and heart



Hepatic and endocrine complications

Symptoms are often dependent on treatment and disease severity.

Diagnosis⁶



BLOOD TESTS INCLUDING A COMPLETE BLOOD COUNT (CBC)



SPECIAL HEMOGLOBIN TESTS



GENETIC TESTS

Patient Population^{4,5}

WORLDWIDE

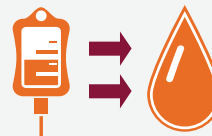
1.5%
(80-90 MILLION)
are **CARRIERS** of the genetic mutation that causes beta-thalassemia

BETA-THALASSEMIA AFFECTS MEN AND WOMEN EQUALLY



Beta-thalassemia is most common in countries around the Mediterranean Sea, the Middle East, Transcaucasia, the Indian subcontinent and the Far East, but has a growing prevalence worldwide

Treatments⁷



STEM CELL TRANSPLANT
Most common for patients **<16 years of age** and/or those with an appropriate match

Supportive Care⁷



BLOOD TRANSFUSIONS

Fe↓

IRON CHELATION THERAPY

B9

FOLIC ACID SUPPLEMENTATION

¹ NIH US National Library of Medicine. Genetics Home Reference: beta thalassemia. Available at: <https://ghr.nlm.nih.gov/condition/beta-thalassemia>. Accessed November 2018.

² National Center for Advancing Translational Sciences, Genetic and Rare Diseases Information Center. Beta-thalassemia. Available at: <http://rarediseases.info.nih.gov/gard/871/beta-thalassemia/resources/1>. Accessed November 2018.

³ Cao A, Galanello R. Beta-thalassemia. Genetics in Medicine. 2010;12:61-76. Available at <http://www.nature.com/gim/journal/v12/n2/full/gim201012a.html?foxtrotcallback=true>. Accessed November 2018.

⁴ Galanello R, Origa R. Beta-thalassemia. Orphanet Journal of Rare Diseases. 2010;5(11). Available at: <https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-5-11>. Accessed November 2018.

⁵ Muncie Jr HL, Campbell J. Alpha and beta thalassemia. Am Fam Physician. 2009; 80(4):339-344. Available at: <https://www.ncbi.nlm.nih.gov/pubmed/19678601>. Accessed November 2018.

⁶ NIH Heart Lung and Blood Institute. How Are Thalassemias Diagnosed? Available at: <https://www.nhlbi.nih.gov/health/health-topics/topics/thalassemia/diagnosis>. Accessed November 2018.

⁷ NIH Heart Lung and Blood Institute. How Are Thalassemias Treated? Available at: <https://www.nhlbi.nih.gov/health/health-topics/topics/thalassemia/treatment>. Accessed November 2018.