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My46 Trait Profile

Beta-thalassemia

Other Names: Cooley's anemia

Beta-thalassemia is genetic blood disorder and is often characterized by chronic anemia (i.e., low blood count). It is caused by mutations in the HBB gene that makes the beta-globin protein.

Characteristics of Beta-thalassemia

Beta-thalassemia is a genetic condition characterized by microcytic hypochromic anemia (i.e., where the red blood cell counts and hemoglobin levels are low) with a varying degree of symptoms from mild (e.g., almost no symptoms) to severe (e.g., anemia appearing within the first two years of life requiring regular blood transfusions). Beta-thalassemia occurs more frequently in individuals of Mediterranean, Middle Eastern, Central Asia, Indian subcontinent and Far Eastern descent.

There are two types of beta thalassemia: beta-thalassemia major and beta-thalassemia intermedia. Symptoms of beta-thalassemia major typically appear between six months and two years of age, and include pallor (i.e., pale appearance) and failure to thrive. If detected early in childhood, regular blood transfusions allow for normal growth and development. However, with frequent blood transfusions, further treatment is often needed to prevent excess iron accumulation. Iron overload can cause health complications during the teenage and young adult years. Beta-thalassemia intermedia results in a spectrum of symptoms ranging from an anemia diagnosed in childhood that may require occasional blood transfusions to no symptoms at all.

Diagnosis/Testing

Most individuals with beta-thalassemia are diagnosed through blood work with specific red blood cell findings suggestive of beta-thalassemia. Diagnosis may also be achieved by genetic testing. Individuals with beta thalassemia have a change or mutation in the HBB gene. This gene makes a beta-globin protein. This protein, along with other globin proteins, makes up a molecule called hemoglobin. Hemoglobin is found in red blood cells and is responsible for carrying oxygen throughout our body. Mutations in the HBB gene cause less hemoglobin to be made, thus less oxygen is delivered to the body, ultimately interfering with the ability of various organs to work properly.

Management/Surveillance

Management of beta-thalassemia includes regular monitoring of red blood cell counts and hemoglobin measurements, with blood transfusions as needed. During childhood, there should be regular monitoring of growth and development. For some individuals with beta-thalassemia major, if there is a well-matched donor, bone marrow transplantation can be done. Bone marrow transplantation effectively cures the condition.

One of the most common complications associated with blood transfusions is iron overload. Iron chelators (i.e., medications that remove excess iron from the body) are often prescribed to prevent or minimize this complication.

Mode of inheritance

Beta-thalassemia is inherited in an autosomal recessive pattern. This means that an individual has to inherit two HBB mutations (i.e., one from each parent) to be affected with beta thalassemia. If both parents are carriers of a HBB

mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with beta-thalassemia.

Risk to family members

Parents of a child with beta-thalassemia are carriers of beta-thalassemia. If a sibling of a child with beta-thalassemia is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of beta-thalassemia.

Special considerations

None

Resources

Cooley's Anemia Foundation
http://www.cooleysanemia.org/
Genetics Home Reference: Beta thalassemia
http://ghr.nlm.nih.gov/condition/beta-thalassemia
KidsHealth: Beta thalassemia
http://kidshealth.org/parent/medical/heart/beta_thalassemia.html

References

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Cao A, Galanello R, Origa R. (Updated 24 January 2013). Beta-Thalassemia. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1426/. Accessed [01/23/2013].
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