BETA THALASSEMIA

There are two copies of the beta globin gene present in an unaffected individual, one on each chromosome 11. Beta plus variants cause decreased beta globin production, and beta null variants cause a complete absese of beta globin production. Clinical findings depend the type of beta globin variant(s) present.

Beta Thalassemia Trait

- One beta globin variant present (beta plus OR beta null)
- Mild anemia, low MCV on CBC
- Mildly elevated HbA2 on Hb Elect

Beta Thalassemia Intermedia

- Usually results from the presence of two beta plus variants
- Lesser clinical severity than thalassemia major
- Onset usually after 2 years of age, up to age 7
- Presentation includes moderate anemia, splenomegaly, moderate to severe hepatomegaly and bony changes
- Transfusions not usually required to survive, but rather to improve quality of life
- Chelation therapy may be required

Beta Thalassemia Major

- Two beta globin variants present (either one beta plus and one beta null or two beta null variants)
- Diagnosis usually made before 2 years of age
- Severe anemia, fatal if untreated
- Splenomegaly
- Secondary iron overload causing organ damage if untreated
- Management includes chronic transfusions, chelation therapy and ongoing monitoring for complications

For more information about about local resources for these conditions, scan the QR code or visit our website

nymacgenetics.org/us-virgin-islands

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Other forms of sickle cell disease can occur when a person has one copy of hemoglobin S variant with another beta globin variant present.

Hemoglobin S-Beta Thalassemia

- One hemoglobin S variant AND one beta thalassemia variant present
- Moderate to severe hemolytic anemia
- Recurrent pain episodes
- Splenomegaly
- Clinical severity depends on the type of beta thalassemia variant inherited
  - Hemoglobin S-beta plus thalassemia tends to be less severe than Hemoglobin S-beta null thalassemia
  - Often difficult to distinguish between sickle cell disease and Hemoglobin S-beta null thalassemia on Hb elect

Hemoglobin C-Beta Thalassemia

- One hemoglobin C variant AND one beta thalassemia variant present
- Clinical severity depends on the type of beta thalassemia variant inherited
- Hemoglobin C-beta plus thalassemia causes a mild anemia with low MCV and 65-70% Hb C, 20-30% Hb A and increased Hb F on Hb elect
- Hemoglobin C-beta null thalassemia causes moderately severe anemia, splenomegaly and possible bone changes with Hb C, no Hb A and increased Hb F on Hb elect

Hemoglobin E-Beta Thalassemia

- One hemoglobin E variant AND one beta thalassemia variant present
- Clinical severity depends on the type of beta thalassemia variant inherited
- Hemoglobin E-beta plus thalassemia causes moderate anemia, splenomegaly, jaundice, microcytosis and the presence of 40% Hb E, 1-30% Hb A and 30-50% Hb F on Hb elect
- Hemoglobin E-beta null thalassemia causes severe anemia, splenomegaly, jaundice, bone marrow expansion, microcytosis and the presence of Hb E with significant increase in Hb F (30 - 60%) and no Hb A on Hb elect. Treatment is similar to beta thalassemia major

These resources are supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling $861,180 with 0 percent financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov.