Beta Thalassemia

β-thalassemia is an inherited microcytic, hypochromic anemia which occurs in Mediterranean populations. These patients have problems synthesizing beta-globin and lead to decreased amounts of the normal hemoglobin tetramer form. One presentation is β-thalassemia minor, where diagnosis occurs with increased HbA2 fraction (> 3.5%), and patients are typically asymptomatic and require no intervention. β-thalassemia major, or Cooley's anemia, is more severe and patients need regular blood transfusions, which may lead to a secondary hemochromatosis. On skull x-ray, these patients show a crew-cut appearance due to marrow expansion.

PLAY PICMONIC

Mechanism

Microcytic, Hypochromic Anemia

Small-cells with Hippo-chrome and Anemone

Lab results in patient with beta thalassemia show a microcytic, hypochromic anemia. This means that the mean corpuscular volume is less than 80 (MCV <80).

Mediterranean Populations

Mediterranean Sea

This particular dyscrasia is prevalent among Mediterranean populations; particularly Greece, Turkey and Italy.

Decreased Beta-Globin

Down-arrow Beta-fish-Globe

Hemoglobin is normally composed of two α-chains and two β-chains. In this disorder, β-globin synthesis does not occur properly. There are varying phenotypes, correlating to if only one or both β-chains are effected.

Beta-Thalassemia Minor

Increased HbA2

Up-arrow He-man-globe with A-Apple and (2) Tutu

Patients with β-thalassemia minor are diagnosed by having an increased HbA2 fraction (>3.5%). This type of hemoglobin is described by having two δ-chains and two β-chains (instead of α).

No Intervention

Broken Intervention-sign

Often, these patients are asymptomatic and are monitored for symptoms.

Beta-thalassemia Major

Blood Transfusions

Blood Transfusion-IV

This is a severe microcytic, hypochromic anemia. Untreated, it causes anemia, splenomegaly and severe bone deformities. It progresses to death before age 20. Treatment consists of periodic blood transfusion; splenectomy if splenomegaly is present, and treatment of transfusion-caused iron overload. Cure is possible by bone marrow transplantation.

Hemochromatosis

He-chrome-man

Patients with this disease can develop a secondary hemochromatosis (due to frequent blood transfusions).

Crew-cut on Skull X-Ray

Crew-cut Skull

Patients with this disease develop erythroid hyperplasia and have marrow expansion. This manifests as a "crew-cut" skull on x-ray and "chipmunk facies."

Diagnosis

Electrophoresis

Electric-fairy

Diagnosis for beta thalassemia minor is confirmed with increased HbA2 on hemoglobin electorphoresis. Alternatively, beta thalassemia major is diagnosed by finding increased HbF on hemoglobin electrophoresis.