SICKLE CELL DISEASE

There are two copies of the beta globin gene present in an unaffected individual, one on each chromosome 11. Hemoglobin S results from a specific variant in the beta globin genes, causing red blood cells form a "sickle" shape when deoxygenated, resulting in clinical consequences.
The type of features will depend on when a person has one or two copies of hemoglobin S variant present, or if other beta globin variants are also present.

Sickle Cell Trait

- One hemoglobin S variant present
- Hemoglobin S seen on Hb Elect
- Minimal clinical issues with normal overall life expectancy
- Episodes of hematuria and increased urinary tract infections possible
- In rare instances, extreme lack of oxygen can cause pain episodes or splenic infarctions

Sickle Cell Disease

- Two hemoglobin S variants present
- Onset in early childhood
- Moderate to severe hemolytic anemia
- Recurrent pain episodes **
- Increased incidence and severity of certain infections
- Tissue infarction leading to organ damage and failure
- Management includes:
  - Accurate, early diagnosis
  - Education / prompt recognition of complications
  - Prevention / treatment of infections
  - Management / aggressive treatment of acute vaso-occlusive events, chronic pain and hemolytic anemia
  - Screening for early signs of organ damage
  - Therapeutic intervention

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

nymacgenetics.org/us-virgin-islands

NYMAC regional genetics network

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 SC Disease

- Two beta globin gene variants present – hemoglobin S and hemoglobin C
- Usually milder than sickle cell disease
- Mild hemolytic anemia
- Occasional infarctive crises
- Splenomegaly
- Increased viscosity of the blood causing:
  - Proliferative retinopathy
  - Painful aseptic necrosis of the femoral head (more than in SS disease)
  - Acute chest syndrome

Hemoglobin S-Beta Thalassemia

- One hemoglobin S variant AND a 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 E-Sickle Cell

- One hemoglobin E variant AND one hemoglobin S variant present
- Mild to moderate hemolytic anemia
- Clinical expression is variable: some patients have no symptoms, whereas others have sickle cell-related complications.
- Less severe as compared to more common forms of sickle cell disease.

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.