

## Genetic Abnormalities Affecting Hemoglobin Structure and Function

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### DESCRIPTION

Hemoglobin, the oxygen-carrying protein in red blood cells, is vital for the transport of oxygen from the lungs to tissues throughout the body. Its intricate structure and function can be profoundly affected by genetic abnormalities, leading to a range of disorders known as hemoglobinopathies. These disorders, often inherited, result from mutations in the genes encoding hemoglobin subunits, altering protein structure and impairing oxygen delivery.

### Hemoglobin structure and function

Normal Adult Hemoglobin (HbA) is a tetramer composed of two  $\alpha$ -globin and two  $\beta$ -globin subunits, each containing a heme group that binds oxygen. This structure is important for its role in reversible oxygen binding and release, facilitated by cooperative interactions between subunits.

### Genetic basis of hemoglobinopathies

Hemoglobinopathies arise from mutations affecting either the  $\alpha$ -globin or  $\beta$ -globin genes. These mutations can lead to structural abnormalities in hemoglobin, affecting its stability, oxygen affinity, or ability to function properly.

### Sickle Cell Disease (SCD)

SCD is caused by a point mutation in the  $\beta$ -globin gene (*HBB*), where glutamic acid is substituted with valine at position 6 ( $\beta^6$  Glu  $\rightarrow$  Val). This results in the production of abnormal hemoglobin known as Hemoglobin S (HbS). Under conditions of low oxygen tension, HbS molecules polymerize, causing red blood cells to deform into a sickle shape. These sickled cells can obstruct blood flow, leading to tissue damage, pain crises, and organ dysfunction.

**Thalassemias:** Thalassemias are characterized by reduced synthesis of  $\alpha$ -globin or  $\beta$ -globin chains due to mutations affecting the corresponding genes (*HBA1/HBA2* for  $\alpha$ -globin and *HBB* for  $\beta$ -globin). Depending on the severity of gene mutations, thalassemias can lead to decreased hemoglobin production, resulting in anemia and related complications.

**Alpha thalassemia:** Deletions or mutations affecting one or more  $\alpha$ -globin genes can lead to impaired  $\alpha$ -globin chain synthesis. Severe forms (deletion of all four  $\alpha$ -globin genes) can cause hydrops fetalis in utero, while milder forms may result in hemolytic anemia.

**Beta thalassemia:** Mutations that decrease or abolish  $\beta$ -globin chain synthesis result in reduced levels of HbA. Depending on the mutation, individuals may exhibit  $\beta$ -thalassemia minor (mild anemia) to  $\beta$ -thalassemia major (severe anemia requiring lifelong transfusions).

### Clinical manifestations and management

The clinical manifestations of hemoglobinopathies vary widely based on the specific mutation and its impact on hemoglobin synthesis and function. Common symptoms include anemia, jaundice, fatigue, and in severe cases, organ damage.

**Diagnosis:** Diagnosis typically involves hemoglobin electrophoresis, which separates different hemoglobin types based on their electrical charge. Molecular genetic testing can identify specific mutations responsible for hemoglobinopathies, aiding in genetic counseling and prenatal diagnosis.

### Treatment and management

Management of hemoglobinopathies focuses on alleviating symptoms, preventing complications, and improving quality of life:

**Transfusion therapy:** Regular blood transfusions are necessary for individuals with severe forms of thalassemia major to maintain adequate hemoglobin levels.

**Hydroxyurea:** This medication increases production of fetal hemoglobin (HbF), which can reduce symptoms and complications in individuals with sickle cell disease.

**Bone marrow transplantation:** Curative in selected cases, bone marrow transplantation can replace defective stem cells with healthy ones capable of producing normal hemoglobin.

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## CONCLUSION

Genetic abnormalities affecting hemoglobin structure and function strengthen a diverse group of disorders known as hemoglobinopathies. These conditions, resulting from mutations in the genes encoding hemoglobin subunits, disrupt oxygen transport and lead to varying degrees of anemia and

other complications. Advances in molecular genetics have improved our understanding of these disorders, enabling better diagnosis, management, and therapeutic interventions to enhance the quality of life for affected individuals. Ongoing research continues to explore new treatments and potential cures for these challenging conditions, provides hope for the future of patients with hemoglobinopathies.