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When the Study of the Globin Genes is Useful?

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ABSTRACT

Hemoglobinopathies constitute a major health problem worldwide, with a high carrier frequency, particularly in certain regions where malaria has been endemic.

These disorders are characterized by a vast clinical and hematological phenotypic heterogeneity.

Based on the gene involved and the type of defect, the hemoglobinopathies can be broadly classified into thalassemias and abnormal structural variants.

Thalassemias are a group of blood genetic disorders, inherited in autosomal recessive manner, caused by reduced or absent synthesis of globin chains.

Defects in these genes can produce abnormal hemoglobin and anemia. Abnormal hemoglobin appears in one of three basic conditions: structural defects in the hemoglobin molecule, diminished production of one of the two subunits of the hemoglobin molecule (thalassemias), and abnormal association of otherwise normal subunits.