HEMOGLOBINOPATHIES The hemoglobinopathies are a group of inherited disorders where there is abnormal production or structure of the hemoglobin molecule.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 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) Severe anemia (fatal if untreated), Secondary iron overload causing organ damage if untreated Splenomegaly, growth delay Management includes chronic transfusions, chelation therapy and ongoing monitoring for complications SICKLE CELL DISEASE There are two copies of the beta globin gene present in an unaffected individual, one on each chromosome 11.Normal CBC & Hb Elect Alpha Thalassemia Trait Two alpha globin gene deleted (CIS form = both genes deleted on the same chromosome; TRANS form one gene deleted from each chromosome) Normal CBC & Hb Elect Hemoglobin H Disease Three alpha globin gene deleted Hb H present on Hb elect Variable clinical presentation Hemoglobin H–Constant Spring Two alpha globin gene deleted in CIS, plus a Constant Spring variant on a third alpha globin gene Hb H present on Hb elect More severe clinical course than Hemoglobin H disease Likely to require transfusions Moderate to severe splenomegaly, growth delay Alpha Thalassemia Major All four alpha globin gene deleted Hb Barts present on Hb elect Survival is possible through intrauterine intervention.There are two main types of hemoglobinopathies: Thalassemia Syndromes Disorders of decreased globin chain production Alpha Thalassemia Beta Thalassemia Hemoglobin Variants Disorders that produce structurally abnormal globin proteins Hemoglobin S, C, E, etc.Alpha Thalassemia There are four copies of the alpha globin gene present in an unaffected individual, two on each chromosome 16 Clinical findings depend on the number of alpha globin genes deleted Silent Carrier One alpha globin gene deleted.Beta Thalassemia There are two copies of the beta globin gene present in an unaffected individual, one on each chromosome 11.Following birth, regular transfusion therapy and chelation .required