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
- Normal CBC
- Normal Hb Elect

Hemoglobin H Disease

- Three alpha globin gene deleted
- Hb H present on Hb elect
- Variable clinical presentation
- Mild to moderate anemia
- May require intermittent transfusions
- Complications can include splenomegaly, growth failure, bony deformities

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 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 with no hematological abnormalities
- Normal Hb Elect

Alpha Thalassemia Major

- Also known as hydrops fettles
- All four alpha globin gene deleted
- Hb Barts present on Hb elect
- Death in utero
- Serious maternal complications can occur
- Survival is possible through intrauterine intervention. Following birth, regular transfusion therapy and chelation required.

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