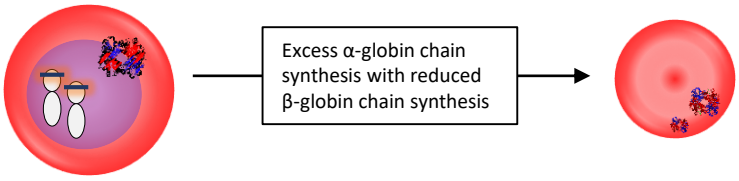
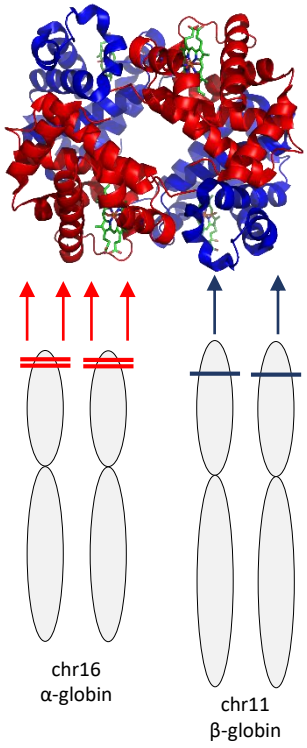
Thalassemia:

- genetic disorder
- *quantitative* abnormalities in α or β hemoglobin chains
- contrast qualitative abn. as in sickle cell anaemia
- clinically heterogeneous because various genetic lesions variably impair globin-chain synthesis, and other genetic modifiers may exist

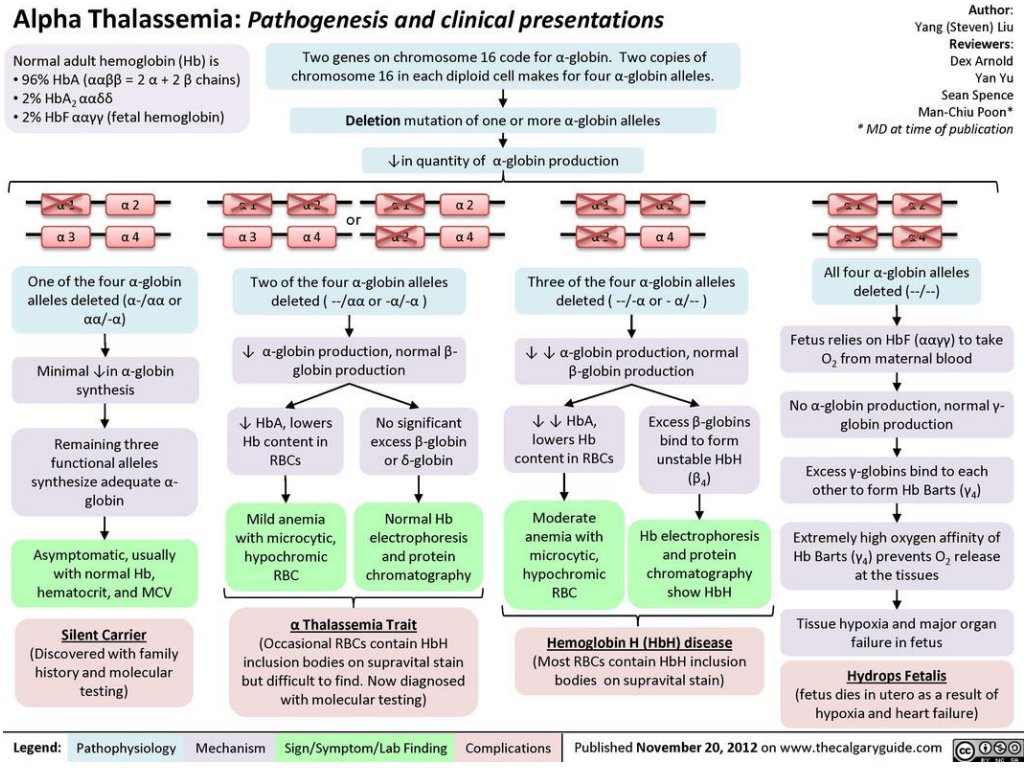


The erythroblast fails to synthesize mutated β-globin. These cells may also undergo apoptosis as unpaired α-chains form hemichromes

Hypochromic, microcytic mature red cells
Target cells are evident
Formation of inclusion bodies and Heinz bodies can also be seen
Precipitation of the unpaired protein causes cell death and cell membrane abnormalities



The α-Thalassemias



The β-Thalassemias

Name	Clinical symptoms	Alleles
Thalassemia minor	Microcytic anemia.	β^+/β β^o/β
Thalassemia intermedia	Mild to moderate anemia; may have slow growth and bone abnormalities.	β^+/β^+ β^o/β^+
Thalassemia major	Severe microcytic, hypochromic anemia. Causes splenomegaly and severe bone deformities. Treatment consists of periodic blood transfusion; splenectomy for splenomegaly.	β^o/β^o

Absence of β -globin called β-zero thalassemia.
Reduced amount of β -globin called β -plus thalassemia.

Having either B0 or B+ thalassemia does not necessarily predict disease severity, however.