Alpha & Beta thalassemia

Alpha

- Chromosome 16
- There are 4 gene mutations!
  - If 1/2 genes mutated → Silent carrier
  - If 3 genes mutated → Hemoglobin H disease!

Extra Beta chains Bind each other in a compound called "HBH"

Extra delta chains Bind each other and called: "HB- Barts"

if 4 genes are mutated: Hydrops fetalis

- The problem in both cases that These compounds have High oxygen affinity So oxygen is Not delivered to tissues!

Beta

- Chromosome 11
- β^0 → No production of beta chains
- β^+ → Decreased Production
- β → Normal production

Okay now lets mix them!!

- β/β^+ → silent or mild thalassemia (thalassemia minor)
- β^+/β^+ → Thalassemia Intermedia
- β^+/β^0 or β^0/β^0 → Thalassemia major (Cooly anemia)

*Note: (Important one!)

- In Beta thalassemia, alpha chains don't bind each other, so the problem here is hemolysis in spleen, not the high oxygen affinity! → and bone marrow

- Thalassemia traits are carriers and usually Asymptomatic!

Symptoms (thalassemia major):

- Histology: - Basophilic stippling of RBCs. - Target cells - Normoblasts
- Clinical: Thalassemia major starts after 6 monthes. (why?)

Because in the first 6 monthes we use fetal Hemoglobin!

- skeletal abnormalities
- Hemochromatosis
- Growth retardation

Diagnosis:

- Hemoglobin electrophoresis test
- In all types of β-thal, there is increase in HgA2 and HgF percentages
- In β-thal major, HgA is absent or markedly decreased
- In HgH disease, HgH and Hg Barts bands appear
- In α-thal carrier and minor, no abnormality is found. Genetic testing is available

Treatment:

- Thalassemia major needs Blood transfusion.
- Thalassemia intermedia and HBH Do Not need blood transfusion