Alpha & Beta thalassemia



Alpha

- Chromosome 16

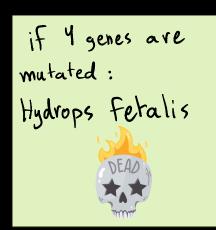
- There are 4 gene mutations!

- IF 1/2 genes mutated - Silent carrier

Lif 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"



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

Beta

- Chromosome 11

- B -> No production of beta chains

- B -> Decreased Production

-B - Normal production

Okay now lets mix them !!

-B/Bt - silent or mild thalassemia (thalassemia

_B/B __ Thalassenia Intermedia

 B^{\dagger}/B° or B^{\prime}/B° 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 Asymptomortic Symptoms (thalassemia major):

- Histology: Basophilic stippling of RBCs. - Target cells - Normoblasts

6 monthes we use

Sclinical: Thalassemia major starts after 6 monthes. (why?)

- steletal abnormalities
- Hemochromatosis
- Growth retardation

Because in the First Fetal Hemoglobin!



- · Hemoglobin electrophoresis test
 - In all types of $\beta\text{-thal}, there is increase in HgA2 and HgF$
- 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

-Thalassemia intermedia and Not need blood transfusion