MUTATIONS

{ وَلَا تَحْسَبَنَّ اللَّهَ غَافِلًا عَمَّا يَعْمَلُ الظَّالِمُونَ إِنَّمَا يُؤَخِّرُهُمْ لِيَوْمٍ تَشْخَصُ فِيهِ الْأَبْصَارُ }

Quantitative :

1] α - Thalassemias : underproduction of α-globin chain

- Silent: 1 of 4 genes is deleted
- Minor (trait): 2 of 4 genes are deleted
- Hb. H disease: 3 of 4 deleted genes with high level of β chains
- Major (Hydrops fetalis): 4 of 4 genes are deleted with fetal hemoglobin of γ chains

2] β-Thalassemias : underproduction of β-globin chain

- Trait : one normal β-globin with one mutated gene "β^0 complete lack of β chain , β^+ some expression β chain"
- Intermedia: Moderate anemia due to partial β globin synthesis defect " $\beta^+\beta^+$ | $\beta^+\beta^0$ | $\beta^E\beta^+$ | $\beta^E\beta^0$ "
- Major (cooley's anemia): complete lack of HbA ⟶ HbA غياب الـ β chains تلقائيا راح يزيد لتعويض الـ

Qualitative :

1] Sickle cell hemoglobin {HbS} : Changing of AA in 6th position of β-globin " Glu(-) ⟶ Val (non polar)
2] Sickle cell trait : Individuals with both HbA (adult) & HbS (sickle)
3] Hemoglobin C {HbC}: Changing of AA in 6th position of β-globin " Glu(-) ⟶ Lys(+)
4] HbSC : Individuals with both β_C & β_S
5] Hemoglobin E : point mutation in codon 26 that changes "Glu(-) ⟶ Lys(+) "
6] Hb Hammersmith : point mutation leading to the change of AA " phe (non polar) ⟶ Ser (polar) "
7] Hb constant spring {HbCS} : Mutation at the termination codon of α_2-globin gene الجين اللي ما بينهم بكون التاكيد على α-globin leading to crest a longer chain
8] Hb Cowtown : Substitution of " His 146(+) ⟶ Leu (non polar) " producing more Hb in the R State
9] Hb Yakima : Substitution of " Asp(-) 99 ⟶ His(+) " leading to the stabilization of the R State R State الـ stabilization التالي بصير عندنا His ما بتعايد الـ Asp كانت الـ interaction راح تروح الـ His ، راح نبدل بسيرة الـ Asp في الـ T State الـ
10] Hb Kansas : Substitution of " Asn 102 ⟶ Thr (non polar) " leading to the stabilization of the T State T State الـ stabilization التالي بصير عندنا His ما بتعايد الـ Asn كانت الـ interaction راح تروح الـ Thr ، راح نبدل بسيرة الـ Asn في الـ R State الـ

Altered Oxygen Transport :

1] Methemoglobin {HbM} : Iron in the heme group is oxidized from ferrous (Fe^{2+}) to ferric (Fe^{3+})
2] Hereditary persistence of fetal hemoglobin {HPFH} : Genetic condition in which the production of fetal hemoglobin ($HbF, \alpha_2\gamma_2$) continues into adulthood instead of switching off after birth

لا تنسوا أهل غزة من الدعاء ، الله يوفقكم ويسهل أموركم جميعا ♥