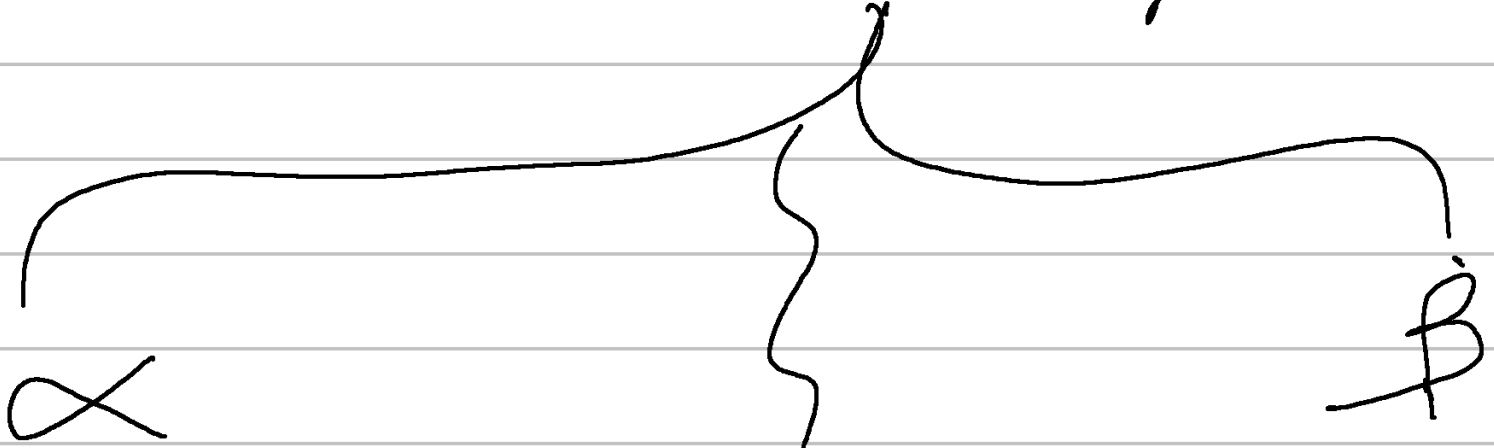
hemoglobinopathies abnormalty locations



β thalassemia: mainly point mut

1) α thalassemia: deletion (mostly) or point mutation

HbS (sickle): 6th amino acid glutamic acid ⇒ valine

2) HbCS (constant spring): α_2 termination codon mutation

HbC: 6th amino acid glutamic acid ⇒ lysine

$\alpha_1 - \beta_2$ contacts mutation

Hb cowtown: His 146 → leucine

HbSC: both C/S mutations

Hb Yakima: Asp G_1 99 → His

hemoglobin E: codon 26 point mutation causing alternative RNa splice (Cframshift) glutamic acid 26 → lysine

Hb Kansas: Asn G_4 102 → Thr

Hb Hammersmith: mostly by internal point mutation phenylalanine → serine

* methemoglobinemia ; methemoglobin reductase AKA (NADH - cytochrome b5 reductase) enzyme deficiency: many causes toxin, drug, mutations

* Hb Boston : distal histidine mutated to tyrosine which oxidize ferrous iron.

* HbM Iwate: proximal histidine mutated to tyrosine.

(These disease effect the heme part)

HPFH : many individuals have large deletions in δ and β coding region of the cluster there is no deletion in fetal globin genes.

وداركم ألما في فلسطين