Thalassemia

Inherited group of disorders in which Mutations in genes Expressing impaired Synthesis of α & β globulin chains

Classification of Thalassemia

Alpha Thalassemia
Beta "
} According to which gene are defective.

- Heterozygotes are asymptomatic
Homozygotes inherit thalassemia genes from Each parent- often have life threatening Clinical manifestations

Alpha Thalassemia:

Due to insufficient synthesis of α haemoglobin chains & an Excess of β chains

4 genes on Chromosome 16 → Required to Produce α Part of Hb.
2 of which from Each parent.

β thalassemia : Due to Insufficient synthesis of β Hb chains & Excess of α Chains
2 genes from Each parent - on 11 chromosome required to produce Beta region of Hb chain.

Etiology : Mutations associated with Thalassemia are passed from parents to children.
Mutation in the DNA cells that make Hb.

Signs & Symptoms :
- Fatigue
- Weakness
- Pale or Yellowish skin.
- Facial bone deformities
- Slow growth
- Abd Swelling
- Dark urine

Pathogenesis :

β thalassemia occurs.

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Quantitative reduction of β globulin chains

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They are caused by Mutations, affecting β globulin locus.

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Then genetic defects lead to a variable reduction in β globulin output ranging from Minimal deficit → to complete absence

(Mild thalassemia) (β-thalassemia)

Excess of Normal α globulin chains which are unaffected

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Result accumulation with in Erythrocyte Precursors

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BCO3 → α chains are not able to form Viable tetramers & instead Precipitate in Red cell Precursors in bone marrow forming Inclusion bodies

which are responsible

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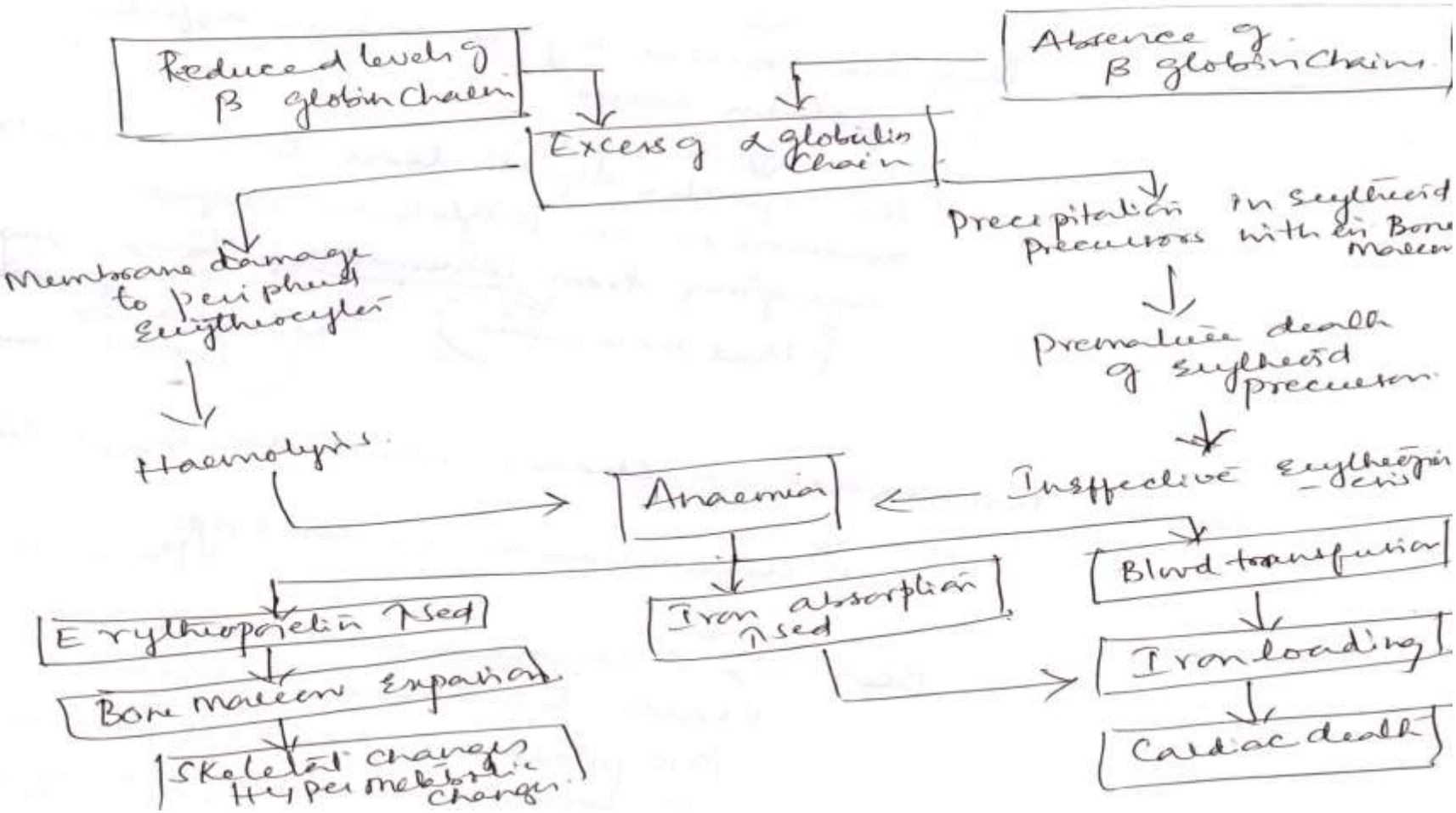
for Extensive destruction of Erythroid precursors

∴ Hence ineffective Erythropoiesis. that ~~underlies~~

Underlies β thalassemia



Treatment :

For Mild Thalassemia :

Signs & symp are Mild & Thalassemia Minor.

Occasionally treatment is Needed, if needed.

- Blood transfusions — particularly after
  - Surgery
  - After having baby
  - or
  - To help manage thalassemia complications

If pat suffer from severe β thalassemia will need Blood transfusion

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Bcoz of this treatment, can cause Iron overload.

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To remove excess Iron oral Medication Deferasirox used to help to remove excess of Iron.

Severe Thalassemia : 1) frequent blood transfusions for every few wks.

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Extra iron can damage liver Heart & other organs.

2) Bone Marrow transplant

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which can eliminate life long blood transfusions & drugs to control Iron overload.

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