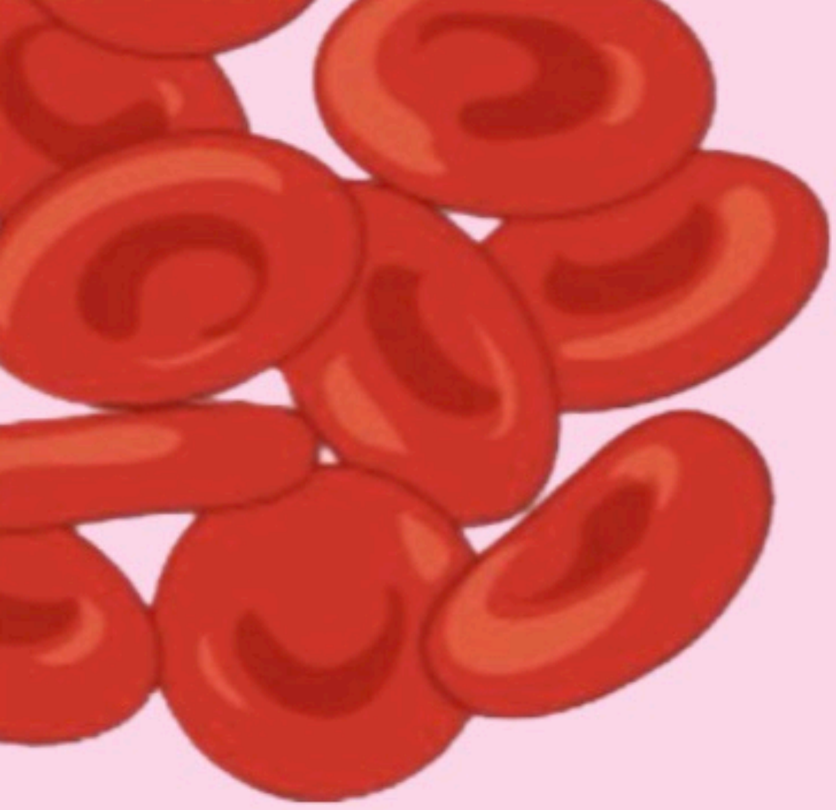


النادي الطبي

HEMATOPOIETIC & LYMPHATIC SYSTEM

-HAYAT BATCH-

SUBJECT : Biochemistry

LEC NO. : 4

DONE BY : Esra'a Khaled

وَقُل رَّبِّ زِدْنِي عِلْمًا

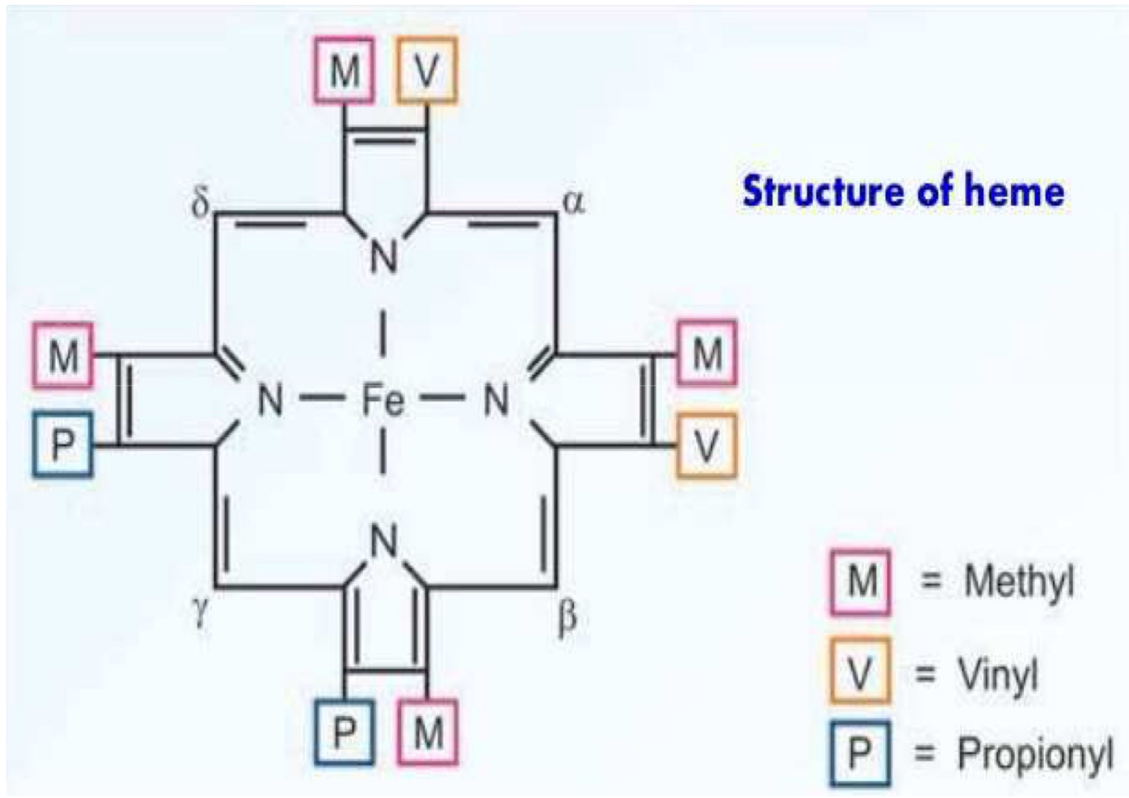
Porphyria

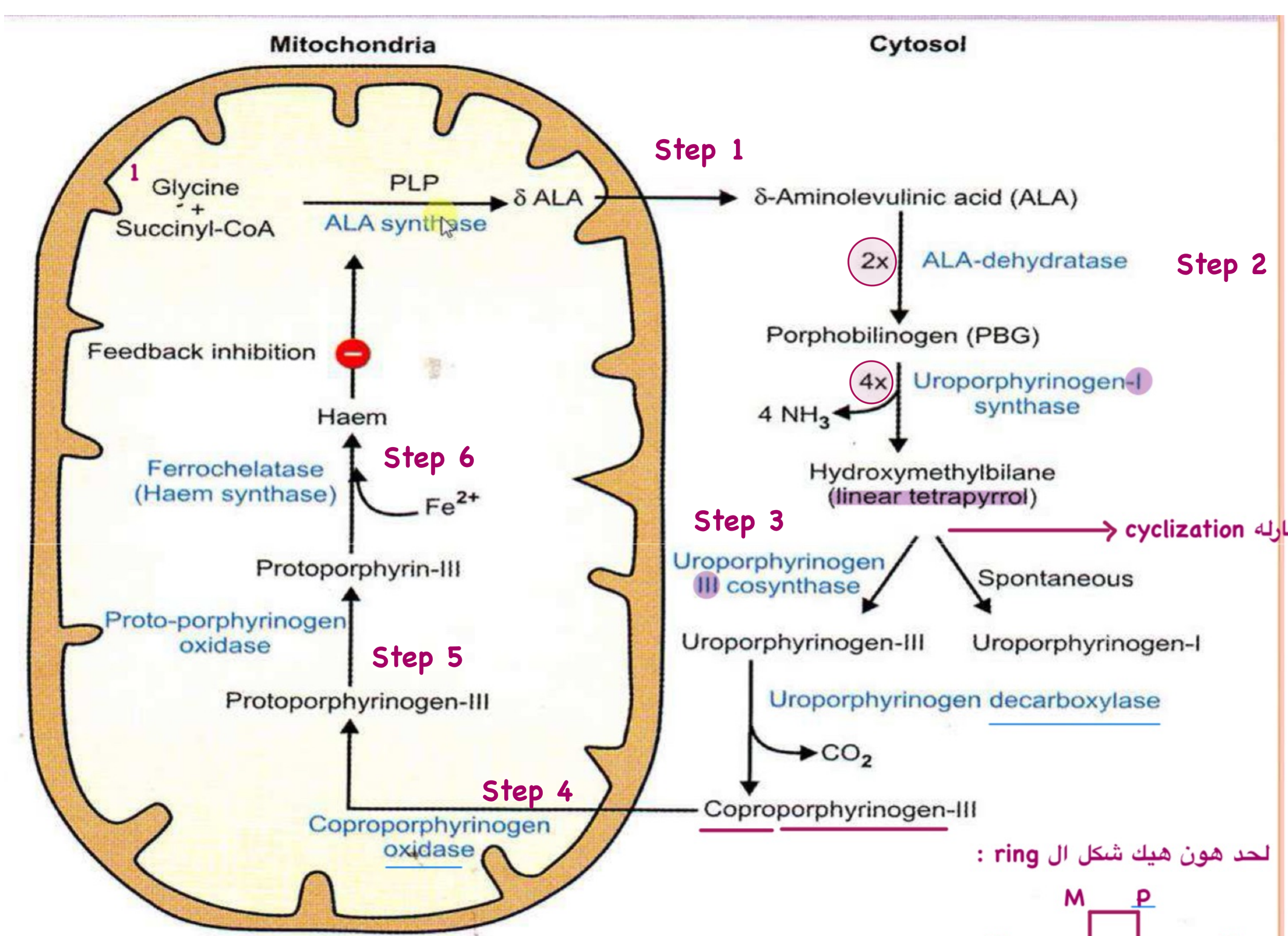
* Porphin ring : cyclic tetrapyrrole united by 4 methene (=CH-) bridges

* Biosynthesis of heme occurs mainly in the liver and bone marrow (both in mitochondria and cytoplasm)

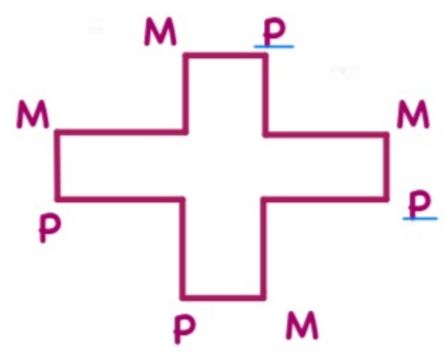
6 Steps :

1. Formation of δ-aminolevulinic acid (ALA)
2. Formation of porphobilinogen (PBG)
3. Formation of uroporphyrinogen III
4. Formation of protoporphyrinogen III
5. Formation of protoporphyrin IX
6. Formation of heme





لحد هون هيك شكل ال ring :



* Porphyria : is a metabolic disease caused by congenital deficiency of one of the enzymes needed for heme synthesis

* The symptoms depend on the site of the defect as following :

1) Enzyme defect before the formation of porphyrinogens : this occure in acute intermittent porphyria due to deficiency of uroporphyrinogen 1 synthase leading to accoumlation of a ALA and porphobilinogen

2) Enzymes defect after the formation of porphyrinogens : this occure in porphyria cutanea tarda due to deficiency of uroporphyrinogen decarboxylase and hereditary copropophyria due to deficiency of coproporphyrinogen oxidase , patient will suffer from 1. photosensitivity 2. skin damage 3. scarring

Hemolytic Anemias

1) Intrinsic causes : defect of RBCs cells membrane as in (hereditary spherocytosis and hereditary elliptocytosis) caused by abnormalities in the amount or structure of spectrin or causes inside the RBCs include (hemoglobinopathies and enzymopathies)

2) Extrinsic causes : portal hypertension in hypersplenism , immunologic abnormalities (SLE , rheumatoid) and prosthetic heart valves recipient

* Laboratory investigations (specific tests) :
  1. Hb electrophoresis (HbS)
  2. Red cell enzyme (G6PD or PK deficiency)
  3. Osmotic fragility (hereditary spherocytosis)
  4. Coombs test

* Reticulocyte count : provides information on the number of relatively immature RBCs in a person's blood sample

* Haptoglobin : When large numbers of RBCs are destroyed , haptoglobin concentrations in the blood will temporarily decrease as the haptoglobin is used up faster than the liver can produce it