

Porphyrias: Introduction



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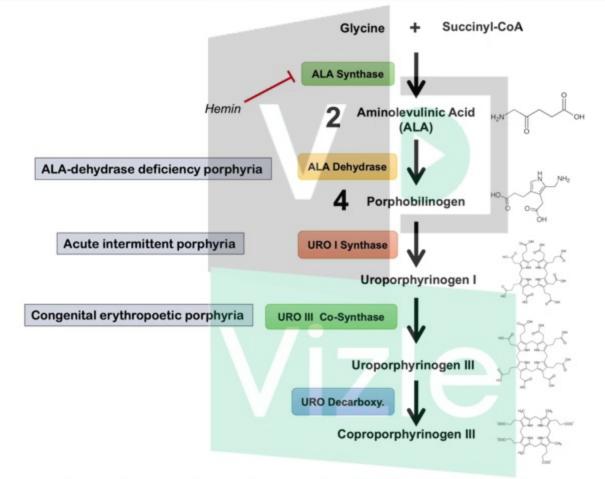
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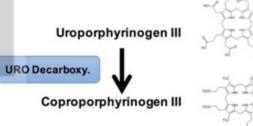






Porphyria Cutanea Tarda (PCT)

- · Most common porphyria condition
- Reduced Activity of Uroporphyrinogen decarboxylase (UROD)
 - · Hepatic enzyme
 - · Due to acquired inhibitor
- Heterozygous UROD mutation predisposes to PCT (Autosomal dominant)





Porphyria Cutanea Tarda (PCT)

- · Accumulation of porphyrinogens
 - Auto-oxidized to photosensitizing porphyrins
- · Blisters, lesions and scars
- · Excessive hair growth
- Tense vesicles/bullae
 - · Sun-exposed skin





- · Diagnosis:
- Urine + 5% HCl Wood Lamp



Acute Intermittent Porphyria (AIP)

- Abdominal
 - Abdominal pain
 - Most common
 - · Vomiting
 - · Constipation/diarrhea
- Neurological
 - Muscle weakness







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