Acute Intermittent Porphyria

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Acute Intermittent Porphyria (short answer question)

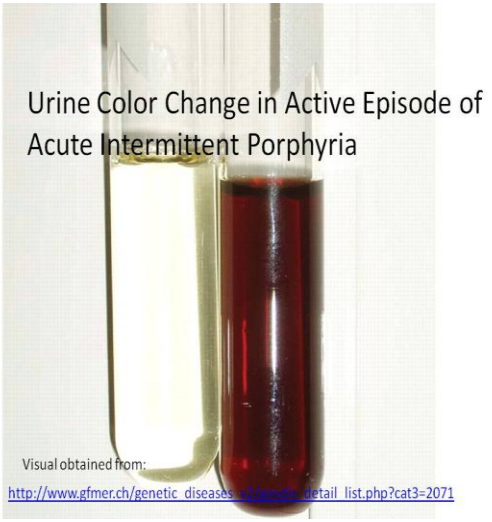
- Most common type,autosomal dominant trait.
- Enzyme defect- Uroporphyrinogen I synthase / HMB synthase/ PBG deaminase.

Reaction blocked: step number 3of heme pathway

- Porphobilinogen  Uroporphyrinogen I

- PBG and ALA levels accumulated in, Blood, tissues and urine.
- Urinary porphobilinogen **photo oxidation**  **Porphobilin**
- ALA Synthase activity increases

- Manifestations:
- Hepatic porphyria and neurological porphyria.
- Symptoms are acute and intermittently.
- Symptoms occur after puberty in humans.(females are more vulnerable)
- Vomiting, constipation, abdominal pain, hypertension.
- Neurological symptoms (due to decrease activity of tryptophan pyrolase.) Because heme concentration decreases. Seizures, anxiety insomnia confusion.
- Tryptophan and 5hydroxytryptamine accumulates.
- no photosensitivity.



- Features of porphyria
- Pain in abdomen
- Polyneuropathy
- Psychological abnormalities
- Purple coloured urine
- Precipitated by medicines (sulphur containing drugs, barbiturates)
- Patients are not photosensitive(enzyme defect prior to formation of uroporphyrinogen
- Treatment:
- By administration of hematin which inhibits the enzyme ALA synthase and accumulation of porphobilinogen