PORPHYRIAS

BIOCHEMISTRY

- Group of disorders either inherited or acquired in the heme synthesis.
- Congenital erythropoietic porphyria- recessive disorder.
- Rest of them are autosomal dominant.
- Mutations are heterogenous.

Clinical Manisfestations

- Erythropeitic and hepatic.
- Hepatic are either acute or chronic.
- Photosensitivity (skin itches and burns)
- These symptoms are due to superoxide radicals, which damage the membranes, and cause the release of lysosomal enzymes.

Acute hepatic porphyrias

1. Acute intermittent porphyria
2. Hereditary coporphyria
3. Varigate porphyria

Symptoms:

- Gastrointestinal
- Neurologic/Psychiatric
- Cardiovascular

- Precipitation of symptoms by:
- Ethanol and barbiturates

Chronic Porphyria

- Porphyria cutanea tarda- the most common porphyria.
- Chronic disease of the liver and erythropoeitic tissue
- Influenced by liver diseases, exposure to sunlight, HIV infections.
- Urine appears pink to red in fluorescent light.

Treatment

- Symptomatic treatment
- Intravenous injection of hemin
- Avoidance of sunlight
- Ingestion of β- carotene

δ-aminolevulinic acid dehydratase

- inhibited by Lead.
- ALA accumulates in the urine.

Acute intermittent porphyria

_Hydroxymethylbilane synthase

- Urine darkens on exposure to light and air.
- Patients are not photosensitive.

Congenital Erythropoietic porphyria

Uroporphyrinogen III synthase

- Patients are photosensitive.

Porphyria Cutanea Tarda

Uroporphyrinogen decarboxylase

- Most common porphyria.
- Patients are photosensitive.

Hereditary Coporphyria

Coporphyrinogens Oxidase

- Acute disease.
- Patients are photosensitive.

Variegate Porphyria

Protoporphyrinogen oxidase

- Acute disease.
- Patients are photosensitive.

Erythropoeitic protoporphyria

Ferrochelatase

- Protoporphyrin acumulate in the bone marrow, RBcs and plasma.
- Patients are photosensitive.

Learning Resources

- Lippincott's Biochemistry