Hepatic porphyrias

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Acute intermittent porphyria (AIP)

The underlying cause is an **AD** hereditary defect of **hydroxymethylbilane synthase** (also known as **porphobilinogen deaminase**, PBGD or uroporphyrinogen I synthase) leading to accumulating hemeprecursors in the liver. It manifests in an **acute attack after exposure** to certain chemicals (steroids, drugs, alcohol), starvation, infection or stress; mostly in the period after puberty. **The main symptoms** are abdominal pain (imitating NPB), constipation, vomiting, hypertension and mental problems (hysteria), headaches, paresis and plegia. There is an increased level of ALA and PBG in the urine. The blood is associated with hyponatremia, hypokalaemia with abnormalities in the metabolism of sugars and fats. The diagnosis is confirmed by decreased activity of PBGD in erythrocytes.

Therapy: in the acute phase = infusion with glucose (inhibits ALA-synthase) and hematin; prevention of another attack is to avoid the triggering factor (certain drugs or alcohol).

Porphyria from 5-aminolevulinic acid dehydratase deficiency (ADP, Doss porphyria)

It is caused by an **AR** hereditary **5-aminolevulinic acid dehydratase** deficiency

Symptoms are abdominal pain and neuropsychic difficulties. ALA and coproporphyrin are present in the urine.

Hereditary coproporphyria (HCP)

It is caused by an **AD** hereditary **coproporphyrinogen oxidase** defect

Symptoms are neuropsychic difficulties, photosensitivity, rarely abdominal pain. Completely **asymptomatic forms** are also common. In the acute phase there is an increased level of ALA, PBG and coproporphyrin (present also in stools) in the urine.

Variegate porphyria (VP)

It is caused by an **AD** hereditary **protoporphyrinogen oxidase** defect.

Symptoms are abdominal pain, neuropsychic difficulties and in some cases even skin symptoms (photosensitivity). Highly increased levels of ALA, PBG, coproporphyrin in the urine with increased excretion of protoporphyrin and coproporphyrin in stools. Template:Netisknout

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