Skin porphyria

Porphyria cutanea tarda (PCT)

It is *'AD'* inherited defect *'of uroporphyrinogendecarboxylase'*, occurring in a ratio of 1:25 000 (the most common form), especially in middle-aged men. Porphyrins are formed in excess in the liver, accumulate there, are transmitted through the bloodstream to the skin, where they cause "photosensitivity", which is a typical symptom. After exposure of the skin to sunlight, large blisters filled with fluid appear, which heal very slowly with the formation of scars and milli (dotted whitish deposits). The skin is hyperpigmented, later atrophic, easily vulnerable. Hypertrichosis occurs on the temples and around the eyes. The clinical manifestation is associated with liver damage caused by alcohol, polyhalogenated hydrocarbons (hexachlorobenzene, dioxin), estrogen therapy, hepatomas, hemochromatosis or hepatitis. Untreated can lead to liver cancer. There is a *'and a non-hereditary form of'* (sporadic, so-called PCT type 1). In the urine we find uroporphyrin, high levels of iron, in 50% of cases high levels liver enzymes.

'Treatment:' repeated venipuncture (300-500 ml at 2-4 week intervals) relieving the body of excess porphyrins and iron + administration of the antimalarial chloroquine (125-250 mg daily), which causes a slow leaching of porphyrins, then sun protection (clothing, special creams) and liver diet.

Congenital (congenital) erythropoietic porphyria (CEP, Günther's disease)

It is an *'AR'* inherited defect of *'uroporphyrinogen-III-synthase (UROS)'* leading to increased porphyrin production in the bone marrow, which accumulates in the body, mainly in erythrocytes. Occurrence is 1: 2−3 mil. This disease usually manifests itself in childhood. *'Manifestations'* of the disease vary - these include dark red urine (due to the presence of uroporphyrin and coproporphyrin), skin sensitivity (blistering, scarring) and darkening, eye sensitivity, eyelash loss, anemia, splenomegaly, red tooth discoloration, excessive hair (especially on the hands and face).

'Treatment:' bone marrow transplantation, sun protection, blood transfusion, splenectomy. Acute photosensitivity reaction in EPP

Protoporphyria (EPP)

It is an *'AD'* inherited defect of *'ferrochelatase'*, which results in the accumulation of protoporphyrin in the liver, bone marrow and skin. The most common "symptoms" are redness, itching and swelling of the skin even after a short (several minutes) exposure of the skin to sunlight. The symptoms disappear after hours to days, with repeated exposure there is scarring of the skin and other variable skin manifestations. The disease usually manifests itself in childhood. In a few percent of cases, liver damage occurs. *'Treatment:'* alleviation with beta-carotene u, antihistamines, melanotan u, phototherapy; prevention is the use of protective clothing and special creams. Unlike acute hepatic porphyrias, EPPs do not exacerbate any drugs. Template:Do not print

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