Cutaneous porphyrias

Porfyria cutanea tarda (PCT)

It is caused by an **AD** hereditary **uroporphyrinogen decarboxylase** defect, occurring in the ratio 1:25 000 (most common form), especially in middle-aged men. Porphyrins are excessively being formed and accumulated in the liver and are transported via the bloodstream to the skin causing **photosensitivity** (typical symptom). After exposure of the skin to sunlight, large blisters filled with fluid start appearing, which heal very slowly with the formation of scars and milla (whitish dotted bumps). The skin is hyperpigmented, later becomes atrophic and 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 treatment, hepatomas, hemochromatosis or hepatitis. Untreated can lead to liver cancer. Uroporphyrin, increased levels of iron and in 50% of cases increased levels of liver enzymes are present in the urine.

A **non-hereditary form** also exists (sporadic, so-called PCT type 1).

Therapy: repeated venipunction (300−500ml in 2−4 weekly intervals) for eliminating excessive porphyrins and iron from the body + antimalarials chloroquine (125-250mg daily), which causes slower secretion of porphyrins + sun protection (clothing, special creams) + liver diet.

Congenital erythropoietic porphyria (CEP, Günther disease)

It is caused by an **AR** hereditary **uroporphyrinogen III synthase** (UROS) defect leading to an increased production of porphyrins in the bone marrow, which accumulate mainly in erythrocytes. The frequency is 1:2−3 mil. It usually manifests itself in childhood.

Symptoms are various: dark red urine (due to uroporphyrin and coproporphyrin), higher sensitivity (blistering, scarring) and darkening of the skin, higher eye sensitivity, loss of eyelashes, anemia, splenomegaly, red tooth color, excessive hair (especially hands and face).

Therapy: bone marrow tranplantation, sun protection, blood transfusion, splenectomy. náhled|Akutní fotosensitivní reakce u EPP

Protoporphyria (EPP)

It is caused by an **AD** hereditary **ferrochelatase** defect leading to an accumulation of protoporphyrin in the liver, bone marrow and skin. It usually manifests itself in childhood. Liver damage occurs in a small percentage of cases.

Symptoms are: redness, itching and swelling of the skin even after a short exposure (minutes) of the skin to sunlight. These symptoms disappear after hours/days but repeated exposure causes scarring of the skin and other variable skin manifestations.

Therapy: alleviation of the manifestations by using beta-carotene, antihistamines, melanotan, phototherapy + sun protection (clothing, special creams). Unlike acute hepatic porphyrias, EPPs are not exacerbated by any drugs. Template:Netisknout [[Category:Pathobiochemistry]