

## Opis choroby \*

### Definicja

A rare disorder of porphyrin and heme metabolism characterized by infantile or childhood onset of severe cutaneous photosensitivity in affected males, presenting as tingling, burning, and itching within minutes of light exposure, often accompanied by swelling and redness of the skin. Pain may persist for hours or days after the initial reaction. Some patients show hepatic involvement and gallstone formation. Laboratory examination reveals increased levels of zinc- and metal-free protoporphyrin. The phenotype in heterozygous females ranges from asymptomatic to severe.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

X-linked dominant erythropoietic protoporphyria

Dominująca protoporfiria erytropoetyczna

sprzężona z chromosomem X

Dominująca protoporfiria sprzężona z chromosomem X

XLDPP

XLP

XLPP

X-linked dominant protoporphyria

XLDPP

XLPP

#### Kod ORPHA

443197

#### Kod OMIM

300752

#### Kod ICD10

E80.0

#### Kod ICD11

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#### \*Źródło

orphanet