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