DISEASE:
X-linked erythropoietic protoporphyria

NAME:	X-linked erythropoietic protoporphyria
DESCRIPTION:	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.
ORPHACODE:	443197
SYNONYMS:	X-linked dominant erythropoietic protoporphyria X-linked dominant protoporphyria XLDPP XLPP
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ALAS2
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2452

RELATED CONTENT

Related Genetic Tests

- Hemochromatosis (17 genes)
- Porphyria (10 genes)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB

Related Analytes

- 5'-aminolevulinate synthase 2

Related Gene Panels

- Hemochromatosis (17 genes) - ULB
- Porphyria (10 genes) - ULB