Niemowlęca cystynoza nefropatyczna
Kod Orpha: 411629 Kod OMIM: 219800

Opis choroby *

Definicja
A subtype of cystinosis characterized by an accumulation of cystine in the organs and tissues, particularly in the kidneys and eyes, and that clinically manifests from infancy with renal Fanconi syndrome, photophobia, hypothyroidism, impaired growth and rickets, in addition to various other systemic effects. Progressive extra-renal manifestations include hypothyroidism, hypogonadism and male infertility, insulin-dependent diabetes, hepatosplenomegaly with portal hypertension, muscle involvement with distal muscle weakness and atrophy, pharyngeal and oral dysfunction, swallowing difficulties, cerebral involvement with hypotonia, speech and walking difficulties, and cerebellar syndrome.
Dane

Klasyfikacja
Podtyp kliniczny

Kod ORPHA
411629

Kod OMIM
219800

Kod ICD10
N16.3*

Kod ICD11
5C60.1

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.