

# 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](#)

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### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie [www.orphanet.pl](http://www.orphanet.pl)