## Opis choroby *

Definicja

A subtype of cystinosis characterized by an accumulation of cystine in different organs and tissues, particularly in the kidneys and eyes, and that clinically manifests between childhood and adolescence with a slowly progressive proximal tubulopathy and/or proteinuria, and photophobia. Extra-renal manifestations (e.g. hypothyroidism, insulin-dependent diabetes, hepatosplenomegaly, muscular and cerebral involvement) are less severe than in the infantile form of the disease.

Dane

**Klasyfikacja**
Podtyp kliniczny

Synonimy
Intermediate cystinosis
Cystynoza młodzieńcza
Cystynoza pośrednia
Juvenile cystinosis

**Kod ORPHA**
411634

**Kod OMIM**
219900

**Kod ICD10**
N16.3*

**Kod ICD11**
5C60.1

---

*Źródło

orphanet