

# Zespół hipotonia - cystynuria

Kod Orpha: 163690 Kod OMIM: 606407

## Opis choroby \*

### Definicja

A rare, genetic disorder of amino acid absorption and transport, characterized by generalized hypotonia at birth, neonatal/infantile failure to thrive (followed by hyperphagia and rapid weight gain in late childhood), cystinuria type 1, nephrolithiasis, growth retardation due to growth hormone deficiency, and minor facial dysmorphism. Dysmorphic features mainly include dolichocephaly and ptosis. Nephrolithiasis occurs at variable ages.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

HCS

HCS

#### Kod ORPHA

163690

#### Kod OMIM

606407

#### Kod ICD10

E72.0

#### Kod ICD11

5C60.Y

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.