

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.