## 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