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