DISEASE:
Hypotonia-cystinuria syndrome

NAME:	Hypotonia-cystinuria syndrome
DESCRIPTION:	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.
ORPHACODE:	163690
SYNONYMS:	HCS
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SLC3A1 PREPL
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RELATED CONTENT

Related Genetic Tests

- Tubulopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- prolyl endopeptidase like
- solute carrier family 3 member 1

Related Gene Panels

- Tubulopathy/Nephrolithiasis (106 genes) - IPG