DISEASE:
2p21 microdeletion syndrome

NAME:	2p21 microdeletion syndrome
DESCRIPTION:	The 2p21 microdeletion syndrome consists of cystinuria, neonatal seizures, hypotonia, severe growthand developmental delay, facial dysmorphism, and lactic acidemia.
ORPHACODE:	163693
SYNONYMS:	2p21 deletion syndrome Del(2)(p21) Monosomy 2p21
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SLC3A1 PREPL CAMKMT PPM1B
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RELATED CONTENT

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- calmodulin-lysine N-methyltransferase
- protein phosphatase, Mg2+/Mn2+ dependent 1B
- prolyl endopeptidase like
- solute carrier family 3 member 1