DISEASE:
Mevalonic aciduria

NAME:	Mevalonic aciduria
DESCRIPTION:	A rare, severe form of mevalonate kinase deficiency (MKD) characterized by dysmorphic features, failure to thrive, psychomotor delay, ocular involvement, hypotonia, progressive ataxia, myopathy, and recurrent inflammatory episodes.
ORPHACODE:	29
SYNONYMS:	Complete mevalonate kinase deficiency MVA
XREF(S):	Orphanet MeSH MedDRA OMIM ICD-10
ANALYTE(S):	MVK
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