DISEASE:
Galactokinase deficiency

NAME:	Galactokinase deficiency
DESCRIPTION:	A rare mild form of galactosemia characterized by early onset of cataract and an absence of the usual signs of classic galactosemia, i.e. feeding difficulties, poor weight gain and growth, lethargy, and jaundice.
ORPHACODE:	79237
SYNONYMS:	GALK deficiency GALK-D Galactokinase deficiency galactosemia Galactosemia type 2
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	GALK1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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