DISEASE:
Oxoglutaric aciduria

NAME:	Oxoglutaric aciduria
DESCRIPTION:	A rare, genetic, inborn error of metabolism disorder characterized by neonatal-onset of developmental delay, hypotonia, hepatomegaly, lactic acidemia, increased creatine kinase levels, elevated alpha-ketoglutaric acid in urine, and a decreased plasma beta-hydroxybutyrate-to-acetoacetate ratio. Pyruvate dehydrogenase deficiency can be associated, leading to hypoglycemia and neurologic anomalies, including seizures.
ORPHACODE:	31
SYNONYMS:	Alpha-ketoglutarate dehydrogenase deficiency
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	OGDH
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