DISEASE:
Hereditary orotic aciduria

NAME:	Hereditary orotic aciduria
DESCRIPTION:	A rare genetic disorder of pyrimidine metabolism characterized by early onset of megaloblastic anemia, global developmental delay, and failure to thrive, associated with massive urinary overexcretion of orotic acid (sometimes with orotic acid crystalluria). Patients without megaloblastic anemia, but with additional manifestations such as epilepsy, have also been reported.
ORPHACODE:	30
SYNONYMS:	Orotidylic decarboxylase deficiency Uridine monophosphate synthetase deficiency
XREF(S):	Orphanet ICD-10 MeSH OMIM MedDRA
ANALYTE(S):	UMPS
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