DISEASE:
3-methylglutaconic aciduria type 8

NAME:	3-methylglutaconic aciduria type 8
DESCRIPTION:	A rare organic aciduria characterized by neonatal onset of hypotonia, recurrent apneic episodes, lack of psychomotor development, feeding difficulties, extrapyramidal signs, and seizures. Other reported features include microcephaly, sensorineural deafness, bradycardia, and neutropenia. Laboratory studies show increased serum lactate and urinary excretion of 3-methylglutaconic acid. Brain imaging may reveal progressive cerebral atrophy. The disease is lethal in infancy.
ORPHACODE:	505208
SYNONYMS:	MGA8
XREF(S):	Orphanet
ANALYTE(S):	HTRA2
CREATED:	13 May 2019 - 01:02
CHANGED:	01 May 2022 - 06:55

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