

DISEASE:
3-methylglutaconic aciduria type 7

NAME:	3-methylglutaconic aciduria type 7
DESCRIPTION:	A rare organic aciduria characterized by increased urinary excretion of 3-methylglutaconic acid, variably associated with neutropenia (sometimes causing recurrent severe infections and potentially resulting in leukemia) and progressive neurologic manifestations, such as global developmental delay, intellectual disability, hypotonia, movement disorder, and seizures. Microcephaly, cataract, facial dysmorphism, growth retardation, endocrine abnormalities, and cardiomyopathy have also been reported. Brain imaging may show cerebral or cerebellar atrophy, or abnormalities of the basal ganglia.
ORPHACODE:	445038
SYNONYMS:	3-methylglutaconic aciduria-cataract-neurologic involvement-neutropenia syndrome MGA7
XREF(S):	Orphanet OMIM ICD-10 OMIM
ANALYTE(S):	CLPB
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