DISEASE:
3-methylglutaconic aciduria type 9

NAME:	3-methylglutaconic aciduria type 9
DESCRIPTION:	A rare organic aciduria characterized by early onset of global developmental delay with severe intellectual disability, seizures, and 3-methylglutaconic aciduria. Additional features are hypotonia, hyperactivity and aggressive behavior, optic atrophy, or spasticity. Brain imaging may show generalized cerebral atrophy and white matter abnormalities.
ORPHACODE:	505216
SYNONYMS:	3-methylglutaconic aciduria-epilepsy-spasticity-severe intellectual disability syndrome MGA9
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TIMM50
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- translocase of inner mitochondrial membrane 50