

**DISEASE:**  
**3-methylglutaconic aciduria type 8**

<b>NAME:</b>	3-methylglutaconic aciduria type 8
<b>DESCRIPTION:</b>	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.
<b>ORPHACODE:</b>	505208
<b>SYNOMYS:</b>	MGA8
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	<a href="#">HTRA2</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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