

**DISEASE:**  
**3-methylglutaconic aciduria type 1**

|                     |                                                                                                                                                                                                                                                 |
|---------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>        | 3-methylglutaconic aciduria type 1                                                                                                                                                                                                              |
| <b>DESCRIPTION:</b> | 3-methylglutaconic aciduria (3-MGA) type I is an inborn error of leucine metabolism with a variable clinical phenotype ranging from mildly delayed speech to psychomotor retardation, coma, failure to thrive, metabolic acidosis and dystonia. |
| <b>ORPHACODE:</b>   | 67046                                                                                                                                                                                                                                           |
| <b>SYNONYMS:</b>    | 3-methylglutaconyl-CoA hydratase deficiency<br>3MG-CoA hydratase deficiency<br>MGA1                                                                                                                                                             |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                 |
| <b>ANALYTE(S):</b>  | <u>AUH</u>                                                                                                                                                                                                                                      |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                             |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                             |

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## RELATED CONTENT

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### Related Analytes

- [AU RNA binding methylglutaconyl-CoA hydratase](#)

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