

**DISEASE:**  
**Intermittent maple syrup urine disease**

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| <b>NAME:</b>        | Intermittent maple syrup urine disease                                                                                                                                                                                                                                                                                                          |
| <b>DESCRIPTION:</b> | Intermittent maple syrup urine disease (intermittent MSUD) is a mild form of MSUD (see this term) where patients (when well) are asymptomatic with normal levels of branched-chain amino acids (BCAAs) but with catabolic stress are at risk of acute decompensation with ketoacidosis, which can lead to cerebral edema and coma if untreated. |
| <b>ORPHACODE:</b>   | 268173                                                                                                                                                                                                                                                                                                                                          |
| <b>SYNONYMS:</b>    | Intermittent BCKD deficiency<br>Intermittent MSUD<br>Intermittent branched-chain alpha-ketoacid dehydrogenase deficiency                                                                                                                                                                                                                        |
| <b>XREF(S):</b>     | <a href="#">Orphanet</a><br><a href="#">OMIM</a><br><a href="#">ICD-10</a>                                                                                                                                                                                                                                                                      |
| <b>ANALYTE(S):</b>  | <a href="#">BCKDHA</a><br><a href="#">BCKDHB</a><br><a href="#">DBT</a>                                                                                                                                                                                                                                                                         |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                             |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                             |

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### Related Genetic Tests

- [Ichthyosis \(gene panel\)](#)

### Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

### Related Analytes

- [branched chain keto acid dehydrogenase E1 subunit alpha](#)
- [branched chain keto acid dehydrogenase E1 subunit beta](#)
- [dihydrolipoamide branched chain transacylase E2](#)

### Related Gene Panels

- [Ichthyosis and erythroderma \(98 genes\) - KUL](#)

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