

DISEASE:
Intermediate maple syrup urine disease

NAME:	Intermediate maple syrup urine disease
DESCRIPTION:	Intermediate maple syrup urine disease (intermediate MSUD) is a milder form of MSUD (see this term) characterized by persistently raised branched-chain amino acids (BCAAs) and ketoacids, but fewer or no acute episodes of decompensation.
ORPHACODE:	268162
SYNONYMS:	Intermediate BCKD deficiency Intermediate MSUD Intermediate branched-chain alpha-ketoacid dehydrogenase deficiency
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u> <u>OMIM</u>
ANALYTE(S):	<u>BCKDHA</u> <u>BCKDHB</u> <u>DBT</u> <u>PPM1K</u>
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14
-----------------	---------------------

Source URL: <http://gentest.healthdata.be/disease/1154>

RELATED CONTENT

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](#)
- [protein phosphatase, Mg²⁺/Mn²⁺ dependent 1K](#)

Related Gene Panels

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

Source URL: <http://gentest.healthdata.be/disease/1154>