

DISEASE:
Classic maple syrup urine disease

NAME:	Classic maple syrup urine disease
DESCRIPTION:	Classic maple syrup urine disease (classic MSUD) is the most severe and probably common form of MSUD (see this term) characterized by a maple syrup odor in the cerumen at birth, poor feeding, lethargy and focal dystonia, followed by progressive encephalopathy and central respiratory failure if untreated.
ORPHACODE:	268145
SYNONYMS:	Classic BCKD deficiency Classic MSUD Classic branched-chain alpha-ketoacid dehydrogenase deficiency Classic branched-chain ketoaciduria
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>BCKDHA</u> <u>BCKDHB</u> <u>DBT</u>
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