DISEASE:
2-methylbutyryl-CoA dehydrogenase deficiency

NAME:	2-methylbutyryl-CoA dehydrogenase deficiency
DESCRIPTION:	A rare organic aciduria characterized by impaired isoleucine degradation with increased plasma or whole blood C5 acylcarnitine levels (typically observed in newborn screening) and increased urinary excretion of N-methylbutyrylglycine. The condition is usually clinically asymptomatic, although patients with muscular hypotonia, developmental delay, and seizures (among others) have been reported.
ORPHACODE:	79157
SYNONYMS:	2-methylbutyric aciduria Developmental delay due to 2-methylbutyryl-CoA dehydrogenase deficiency SBCAD deficiency Short/branched-chain acyl-coA dehydrogenase deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ACADSB
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