DISEASE:
Pyruvate dehydrogenase E3 deficiency

NAME:	Pyruvate dehydrogenase E3 deficiency
DESCRIPTION:	Pyruvate dehydrogenase E3 deficiency is a very rare subtype of pyruvate dehydrogenase deficiency (PDHD, see this term) characterized by either early-onset lactic acidosis and delayed development, later-onset neurological dysfunction or liver disease.
ORPHACODE:	2394
SYNONYMS:	DLD deficiency Dihydrolipoamide dehydrogenase deficiency E3-deficient maple syrup urine disease
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DLD
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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