DISEASE:
Pyruvate dehydrogenase E1-alpha deficiency

NAME:	Pyruvate dehydrogenase E1-alpha deficiency
DESCRIPTION:	A disorder that is the most frequent form of pyruvate dehydrogenase deficiency (PDHD) characterized by variable lactic acidosis, impaired psychomotor development, hypotonia and neurological dysfunction.
ORPHACODE:	79243
SYNONYMS:	PDHAD Pyruvate decarboxylase deficiency Pyruvate dehydrogenase complex E1 component subunit alpha deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PDHA1 LONP1
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