Pyruvate dehydrogenase E2 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pyruvate dehydrogenase E2 deficiency. ORPHA:79244

Pyruvate dehydrogenase E2 deficiency is a very rare form of pyruvate dehydrogenase deficiency (PDHD, see this term) characterized by variable lactic acidosis and neurological dysfunction, mainly appearing during childhood.