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# Pyruvate dehydrogenase E1-beta deficiency

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pyruvate dehydrogenase E1-beta deficiency. ORPHA:255138*

Pyruvate dehydrogenase E1-beta deficiency is an extremely rare form of pyruvate dehydrogenase deficiency (PDHD, see this term) characterized by severe lactic acidosis, developmental delay and hypotonia.