Mitochondrial myopathy-lactic acidosis-deafness syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mitochondrial myopathy-lactic acidosis-deafness syndrome. ORPHA:2597

Mitochondrial myopathy-lactic acidosis-deafness is a type of metabolic myopathy described only in two sisters to date, presenting during childhood, and characterized clinically by growth failure, severe muscle weakness, and moderate sensorineural deafness and biochemically by metabolic acidosis, elevated serum pyruvate concentration, hyperalaninemia and hyperalaninuria. There have been no further descriptions in the literature since 1973.