Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria. ORPHA:1933

Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria is characterised by the association of a mitochondrial encephalomyopathy and an aminoacidopathy. It has been described in two brothers presenting with developmental delay, neurological signs, deafness, exercise intolerance, lactic acidosis and elevation of several plasmatic amino acids. Mitochondria morphology was found to be abnormal on muscle biopsy. Transmission is likely to be linked to maternal mitochondrial DNA.