Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency. ORPHA:314637

Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency is a rare mitochondrial oxidative phosphorylation disorder with complex I and IV deficiency characterized by lactic acidosis, hypotonia, hypertrophic cardiomyopathy and global developmental delay. Other clinical features include feeding difficulties, failure to thrive, seizures, optic atrophy and ataxia.