Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation. ORPHA:324525

Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation is a mitochondrial oxidative phosphorylation disorder characterized by hypertrophic and dilated cardiomyopathy, failure to thrive, myopathy with generalized hypotonia and increased creatine kinase, developmental delay and/or regression with cerebral atrophy on brain MRI, renal manifestations including chronic renal failure, renal tubular acidosis and lactic acidosis. Additional clinical features include seizures and respiratory failure.