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Deafness-encephaloneuropathy-obesity-valvulopathy syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Deafness-encephaloneuropathy-obesity-valvulopathy syndrome. ORPHA:254898*

Deafness-encephaloneuropathy-obesity-valvulopathy syndrome is a rare mitochondrial disease with marked clinical variability typically characterized by encephalomyopathy, kidney disease (nephrotic syndrome), optic atrophy, early-onset deafness, pancytopenia, obesity, and cardiac disease (valvulopathy). Additionally, macrocephaly, intellectual disability, hyperlactatemia, elevated lactate/pyruvate ratio, insulin-dependent diabetes, livedo reticularis, liver dysfunction and seizures have also been associated.