

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

Hearing loss-encephaloneuropathy-obesity-valvulopathy syndrome

Utrata słuchu - encefaloneuropatia - otyłość - walwulopatia

Kod ORPHA

254898

Kod OMIM

614651

Kod ICD10

E88.8

Kod ICD11

5C53.22

*Źródło

orphanet