

## 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

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#### \*Źródło

orphanet