## Opis choroby *

Definicja

A rare mitochondrial disease characterized by a variable phenotype comprising congenital sensorineural deafness, intermittent or persistent hypoglycemia, and hepatic and renal dysfunction potentially progressing to organ failure. Serum lactate levels are variably increased, deficiency of mitochondrial respiratory chain complexes I, III, and IV is observed in the liver and in fibroblasts.

Dane

**Klasyfikacja**
Choroba

Synonimy
Syndromic sensorineural deafness due to COXPD
Syndromiczna czuciowo-nerwowa głuchota spowodowana COXPD
Syndromiczna czuciowo-nerwowa utrata słuchu spowodowana COXPD
Syndromic sensorineural hearing loss due to COXPD

**Kod ORPHA**
457223

**Kod OMIM**
617872

**Kod ICD10**
E88.8

**Kod ICD11**
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*Źródło

orphanet