## Opis choroby \*

## Definicja

A rare mitochondrial disease characterized by prenatal or early infantile onset of severe cardiomyopathy, failure to thrive and global developmental delay, sensorineural hearing loss, and severe lactic acidosis. Hepatic involvement and adrenal insufficiency, as well as encephalopathy and anomalies of deep gray matter structures on brain MRI have also been reported.

Dane

Klasyfikacja Synonimy

Choroba QRSL1-related COXPD

QRSL1-related COXPD

Kod ORPHA Kod OMIM Kod ICD10

570491 618835 E88.8

**Kod ICD11** 5C53.23

## \*Źródło

orphanet