

## Opis choroby \*

### Definicja

A rare, genetic, syndromic intellectual disability disease characterized by severe intrauterine and post-natal growth delay, moderate to severe intellectual disability, and neonatal-onset hepatopathy with fibrosis, steatosis, and/or cholestasis, occasionally leading to liver failure. Additional variable manifestations include muscular hypotonia, zinc deficiency, recurrent infections, diabetes mellitus, joint contractures, skin and joint laxity, hypervitaminosis D, and sensorineural hearing loss.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

541423

#### Kod OMIM

617093

#### Kod ICD10

K76.8

#### Kod ICD11

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### \*Źródło

orphanet