## 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

K76.8

**Kod ICD11** 

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\*Źródło

orphanet