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