Opis choroby *

Definicja

A rare peroxisome biogenesis disorder (the most severe variant of Peroxisome biogenesis disorder spectrum) characterized by neuronal migration defects in the brain, dysmorphic craniofacial features, profound hypotonia, neonatal seizures, and liver dysfunction.

Dane

Klasyfikacja
Choroba

Synonimy
Cerebrohepatorenal syndrome
Zespół mózgowo-wątrobowo-nerkowy
ZS
Severe PBD-ZSD
Severe peroxisome biogenesis disorder-Zellweger spectrum disorder
ZS

Kod ORPHA
912

Kod OMIM
614887

Kod ICD10
Q87.8

Kod ICD11
5C57.0

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*Źródło

orphanet