

Opis choroby *

Definicja

Peroxisome biogenesis disorders, Zellweger syndrome spectrum (PBD-ZSS) is a group of autosomal recessive disorders affecting the formation of functional peroxisomes, characterized by sensorineural hearing loss, pigmentary retinal degeneration, multiple organ dysfunction and psychomotor impairment, and is comprised of the phenotypic variants Zellweger syndrome (ZS), neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD) (see these terms).

Dane

Klasyfikacja	Synonimy
Grupa fenomenów	PBD-ZSD Spektrum zaburzenia biogenezy peroksysomów Peroxisome biogenesis disorder spectrum Peroxisome biogenesis disorder-Zellweger spectrum disorder

Kod ORPHA	Kod OMIM	Kod ICD10
79189	202370	Q87.8

Kod ICD11
5C57.0

*Źródło

orphanet