

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

Grupa fenomenów

Synonimy

PBD-ZSD

Spektrum zaburzenia biogenezy peroksysomów

Peroxisome biogenesis disorder spectrum

Peroxisome biogenesis disorder-Zellweger spectrum disorder

Kod ORPHA

79189

Kod OMIM

202370

Kod ICD10

Q87.8

Kod ICD11

5C57.0

*Źródło

orphanet