

Opis choroby *

Definicja

Infantile Refsum disease (IRD) is the mildest variant of the peroxisome biogenesis disorders, Zellweger syndrome spectrum (PBD- ZSS; see this term), characterized by hypotonia, retinitis pigmentosa, developmental delay, sensorineural hearing loss and liver dysfunction. Phenotypic overlap is seen between IRD and neonatal adrenoleukodystrophy (NALD) (see this term).

Dane

Klasyfikacja	Synonimy
Choroba	IRD
	IRD
	Mild PBD-ZSD
	Mild peroxisome biogenesis disorder-Zellweger spectrum disorder

Kod ORPHA	Kod OMIM	Kod ICD10
772	617370	G60.1

Kod ICD11
5C57.1

*[Źródło](#)

orphanet