

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

Choroba

Synonimy

IRD

IRD

Mild PBD-ZSD

Mild peroxisome biogenesis disorder-Zellweger spectrum disorder

Kod ORPHA

772

Kod OMIM

617370

Kod ICD10

G60.1

Kod ICD11

5C57.1

*Źródło

orphanet