

## 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

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#### \*Źródło

orphanet