

Opis choroby *

Definicja

A variant of intermediate severity of the PBD-Zellweger syndrome spectrum (PBD-ZSS) characterized by hypotonia, leukodystrophy, and vision and sensorineural hearing deficiencies. Phenotypic overlap is seen between NALD and infantile Refsum disease (IRD).

Dane

Klasyfikacja

Choroba

Synonimy

Intermediate PBD-ZSD

NALD

Intermediate peroxisome biogenesis disorder-

Zellweger spectrum disorder

NALD

Kod ORPHA

44

Kod OMIM

617370

Kod ICD10

E71.3

Kod ICD11

5A74.Y

*Źródło

orphanet