Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability characterized by global developmental delay, postnatal microcephaly, intellectual disability, ataxia, sensorineural hearing loss, and exocrine pancreatic insufficiency. More variable manifestations include hypotonia, growth retardation, peripheral demyelinating neuropathy, dysmorphic facial features, and additional endocrine abnormalities. Brain imaging may show progressive cerebellar atrophy in some patients.

Dane

Klasyfikacja
Choroba

Synonimy
IMNEPD
IMNEPD

Kod ORPHA
456312

Kod OMIM
616263

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet