

Opis choroby *

Definicja

Intellectual disability-obesity-brain malformations-facial dysmorphism syndrome is a rare, syndromic intellectual disability primarily characterized by moderate to severe intellectual disability, true-to-relative microcephaly and brain abnormalities including a thin corpus callosum, cerebellar hypoplasia, cerebral white matter hypoplasia and multi-focal hyperintensity of cerebral white matter on MRI. Obesity and distinctive craniofacial dysmorphism (including brachycephaly, round face, straight eyebrows, synophrys, hypertelorism, epicanthus, wide and depressed nasal bridge, protruding ears with uplifted lobe, downslanting corners of the mouth) are additional features.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive intellectual disability due to TRAPPC9 deficiency

Autosomalna recesywna niepełnosprawność intelektualna z powodu niedoboru TRAPPC9

Kod ORPHA

352530

Kod OMIM

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Kod ICD10

Q04.8

Kod ICD11

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*Źródło

orphanet