

Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by variable intellectual deficit, macrocephaly, short stature, and facial dysmorphism (such as prominent forehead, prominent supraorbital ridges, hypertelorism, downslanting palpebral fissures, broad nasal tip, anteverted nostrils, thick lower lip, and localized microdontia). Additional reported features include seizures, post-pubertal macroorchidism, obesity, and short, broad hands with tapered fingers.

Dane

Klasyfikacja

Zespół wad wrodzonych X-linked intellectual disability, Atkin type

Synonimy

Niepełnosprawność intelektualna sprzężona z chromosomem X, typu Atkina

Kod ORPHA

1193

Kod OMIM

300431

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet