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

Synonimy
X-linked intellectual disability, Atkin type
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