Opis choroby *

Definicja

An X-linked syndromic intellectual disability characterized by developmental delay, intellectual disability (ID) with severe speech impairment, and short stature. Variable additional clinical features have been associated, including behavioral disturbances, gait abnormalities, tremor, seizures, hypogonadism, truncal obesity, unspecific facial dysmorphism, and small hands and feet.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cabezas syndrome
Niepełnosprawność intelektualna sprzężona z chromosomem X, typ Cabezasa

Kod ORPHA
85293

Kod OMIM
300354

Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet