

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

*Źródło

orphanet