Opis choroby *

Definicja

A rare, X-linked syndromic intellectual disability disorder characterized by mild to moderate intellectual disability, obesity, hypogonadism, tapering fingers and microphallus with small or undescended testes, localized to Xp11.3-Xq23. Additional variable manifestations include alopecia, dental and eyesight anomalies, speech disabilities, and decreased body strength.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MRXS7
MRXS7
Niepełnosprawność intelektualna sprzężona z chromosomem X, typ Ahmada
X-linked intellectual disability, Ahmad type

Kod ORPHA
85274

Kod OMIM
300218

Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet