

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 MRXS7

Synonimy

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

*Źródło

orphanet