Opis choroby *

Definicja

A rare genetic disease associated with mild to severe intellectual deficit that may be associated with behavioral disorders and characteristic physical features including a high forehead, prominent and large ears, hyperextensible finger joints, flat feet with pronation and, in adolescent and adult males, macroorchidism.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
FRAXA syndrome
FXS
Zespół FraX
Zespół FRAXA
Zespół Martina i Bell
FXS
FraX syndrome
Martin-Bell syndrome

Kod ORPHA
908

Kod OMIM
311360, 300624

Kod ICD10
Q99.2

Kod ICD11
LD55

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*Źródło

orphanet