Opis choroby *

Definicja

X-linked intellectual disability, Stoll type is characterised by intellectual deficit, short stature and characteristic facies (hypertelorism, prominent forehead, frontal bossing, a broad nasal tip and anteverted nares). It has been described in four males from three generations of the same family. Two females from this family also displayed intellectual deficit and the characteristic facies. Transmission is X-linked.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
85326

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet