

Opis choroby *

Definicja

X-linked intellectual disability, Shashi type is characterised by moderate intellectual deficit, obesity, macroorchidism and a characteristic facies (large ears, a prominent lower lip and puffy eyelids). It has been described in nine boys from two families. Transmission is X-linked and the causative gene has been localised to the q21.3-q27 region of the X chromosome.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Syndromic X-linked intellectual disability type 11
Syndromiczna niepełnosprawność intelektualna
sprzężona z chromosomem X typu 11

Kod ORPHA

85286

Kod OMIM

300238

Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet