

Opis choroby *

Definicja

X-linked intellectual disability, Siderius type is characterised by mild to borderline intellectual deficit associated with cleft lip/palate. Preaxial polydactyly, large hands and cryptorchidism are sometimes present. The syndrome has been described in seven boys from two families. Transmission is X-linked and the syndrome is caused by mutations in the *PHF8* gene, localised to the p11.21 region of the X chromosome.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85287

Kod OMIM

300263

Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet