

## 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

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### \*Źródło

orphanet