

## Opis choroby \*

### Definicja

Xp22.13p22.2 duplication syndrome is a rare syndromic intellectual disability characterized by developmental delay and intellectual disability, learning and behavioral problems, short stature, thin and sparse hair, mild dysmorphic features, tapering fingers and later onset of scoliosis, obesity and cardiovascular problems (cardiomegaly and cardiomyopathy). Females have normal intelligence.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Dup(X)(p22)  
Dup(X)(p22)  
Dup(X)(p22.13p22.2)  
Duplikacja Xp22  
Dup(X)(p22.13p22.2)  
Duplication Xp22

#### Kod ORPHA

284180

#### Kod OMIM

-

#### Kod ICD10

Q99.8

#### Kod ICD11

LD51

---

#### \*Źródło

orphanet