

## Opis choroby \*

### Definicja

X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome is a rare X-linked syndromic intellectual disability disorder characterized by profound intellectual disability, global developmental delay with absent speech, seizures, large joint contractures, abnormal position of thumbs and middle-age onset of cardiomegaly and atrioventricular valve abnormalities, resulting in subsequent congestive heart failure. Additional features include variable facial dysmorphism (notably large ears with overfolded helix) and large testes.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

324410

#### Kod OMIM

300886

#### Kod ICD10

Q87.8

#### Kod ICD11

LD90

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### \*Źródło

orphanet