

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

*Źródło

orphanet