Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by global development delay, postnatal growth retardation leading to short stature, facial dysmorphism, short hands with tapering fingers and progressive skeletal abnormalities including kyphoscoliosis and pectus carinatum/excavatum. Intellectual disability ranges from mild to severe.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CLS
CLS

Kod ORPHA
192

Kod OMIM
303600

Kod ICD10
Q87.0

Kod ICD11
LD2F.1Y

---

*Źródło

orphanet