

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 | Synonimy |
| Zespół wad wrodzonych CLS | CLS |
| | CLS |

| | | |
|------------------|-----------------|------------------|
| Kod ORPHA | Kod OMIM | Kod ICD10 |
| 192 | 303600 | Q87.0 |

Kod ICD11
LD2F.1Y

*Źródło

orphanet