

## Opis choroby \*

### Definicja

A rare, highly variable, multisystemic disorder mainly characterized by short stature, distinctive facial features, congenital heart defects, cardiomyopathy and an increased risk to develop tumors in childhood.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

648

#### Kod OMIM

616564

#### Kod ICD10

Q87.1

#### Kod ICD11

LD2F.15

---

### \*Źródło

orphanet