

## **Opis choroby \***

### Definicja

A rare multiple congenital anomalies syndrome characterized by short stature, distinctive facial dysmorphism, brachydactyly, stiff and thick skin, muscular pseudohypertrophy, restricted joint mobility, hearing loss, and variable intellectual disability. Cardiovascular and respiratory involvement are common.

### Dane

#### **Klasyfikacja**

Zespół wad wrodzonych Facial dysmorphism-intellectual disability-short

stature-deafness syndrome

Dysmorphia twarzy - niepełnosprawność

intelektualna - niski wzrost - utrata słuchu

Facial dysmorphism-intellectual disability-short

stature-hearing loss syndrome

#### **Kod ORPHA**

2588

#### **Kod OMIM**

139210

#### **Kod ICD10**

Q87.8

#### **Kod ICD11**

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#### \*Źródło

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