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

Synonimy
Facial dysmorphism-intellectual disability-short stature-deafness syndrome
Dysmorfia 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

orphanet