

Opis choroby *

Definicja

Short stature-valvular heart disease-characteristic facies syndrome is characterised by severe short stature with disproportionately short legs, small hands, clinodactyly, valvular heart disease and dysmorphism (ptosis, high-arched palate, abnormal dentition). It has been described in a mother and two daughters. This syndrome is probably transmitted as an autosomal dominant trait.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2868

Kod OMIM

126190

Kod ICD10

Q87.1

Kod ICD11

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*Źródło

orphanet