

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by skeletal dysplasia (including coronal clefting of the vertebral bodies and short limbs and variable congenital heart malformations, such as atrial and ventricular septal defects, right ventricular hypoplasia, and valve defects). There have been no further descriptions in the literature since 1990.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

1354

Kod OMIM

212135

Kod ICD10

Q87.2

Kod ICD11

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*Źródło

orphanet