

Opis choroby *

Definicja

Dysmorphism-pectus carinatum-joint laxity syndrome is characterised by joint laxity, pectus carinatum and facial dysmorphism (mild frontal bossing, a beaked nose with a low nasal bridge, malar hypoplasia, chubby cheeks, a striking philtrum and arched upper lips). It has been described in two siblings. The mode of transmission is unknown.

Dane

Klasyfikacja

Zespół wad wrodzonych Guízar Vázquez-Sánchez-Manzano syndrome
Zespół Guizar Vazqueza, Sancheza i Manzano

Kod ORPHA

2104

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet