

Dysmorfia twarzy - shawl scrotum - luźność stawów

Kod Orpha: 1778 Kod OMIM:

Opis choroby *

Definicja

Facial dysmorphism-shawl scrotum-joint laxity syndrome is characterised by facial dysmorphism (hypertelorism, telecanthus, downslanting palpebral fissures, ptosis, malar hypoplasia, broad nasal bridge, thin upper lip, smooth philtrum, and low-set prominent ears) and associated with joint anomalies (genu valgum or cubitus valgus, hyper-extensible joints, etc.). It has been described in two patients (a mother and her son). The boy also had hypoplastic shawl scrotum and cryptorchidism, and the mother had mild intellectual deficit.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Seaver-Cassidy syndrome
Zespół Seavera i Cassidy

Kod ORPHA

1778

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.