

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	Synonimy
Zespół wad wrodzonych	Seaver-Cassidy syndrome Zespół Seavera i Cassidy

Kod ORPHA	Kod OMIM	Kod ICD10
1778	-	Q87.8

Kod ICD11
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*Źródło

orphanet