## 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