Facial dysmorphism-shawl scrotum-joint laxity syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Facial dysmorphism-shawl scrotum-joint laxity syndrome. ORPHA:1778

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.