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Pterygium colli-intellectual disability-digital anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pterygium colli-intellectual disability-digital anomalies syndrome. ORPHA:2988

Pterygium colli-intellectual disability-digital anomalies syndrome is characterized by pterygium colli, digital anomalies (abnormal small thumbs, widened interphalangeal joints, and broad terminal phalanges), and craniofacial abnormalities (brachycephaly, epicanthic folds, angulated eyebrows, upward slanting of the palpebral fissures, ptosis, hypertelorism, and prominent low-set, posteriorly rotated ears). It has been described in a woman and her son, but the manifestations were much less severe in the mother. The son also had intellectual deficit. The inheritance is either X-linked dominant or autosomal dominant.