Craniofrontonasal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Craniofrontonasal dysplasia. ORPHA:1520

Craniofrontonasal dysplasia is an X-linked malformation syndrome characterized by facial asymmetry (particularly orbital), body asymmetry, midline defects (hypertelorism, frontal bossing, broad grooved or bifid nasal tip, cleft lip and/or palate, high arched palate), skeletal anomalies (clavicle pseudoarthrosis, coronal craniosynostosis, various digital and limb anomalies including syndactyly, clinodactyly of the 5th finger, broad thumbs) and ectodermal dysplasias (dental anomalies, grooved nails, wiry hair). Contrary to most X-linked disorders, females are much more severely affected whereas males are asymptomatic or present with a mild phenotype, frequently only displaying hypertelorism.