Distal trisomy 17q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 17q. ORPHA:3379

Distal trisomy 17q is a rare chromosomal anomaly syndrome with variable phenotype principally characterized by intellectual disability, developmental delay, short stature, craniofacial dysmorphism (incl. microcephaly, low posterior hairline, frontal bossing, bitemporal narrowing, low-set and malformed ears, flat nasal bridge, long philtrum, wide mouth with downturned corners, thin upper lip) and a short, webbed neck, as well as skeletal anomalies (e.g. brachyrhizomelia, poly-/syndactyly) and joint hyperlaxity. Cardiac, cerebral, and urogenital anomalies are also frequently associated.