Distal trisomy 6q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 6q. ORPHA:96098

Distal trisomy 6q is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 6, with highly variable phenotype, typically characterized by growth and developmental delay, intellectual disability, craniofacial dysmorphism (microcephaly, flat facial profile, frontal bossing, hypertelorism, downward-slanting palpebral fissures, flat nasal bridge, anteverted nares, bow shaped mouth, micrognathia), short webbed neck and joint contractures. Cardiac, urogenital, ophthalmologic and hand and foot anomalies, as well as umbilical hernia, spasticity, and seizures, are other features that have been reported.