Distal trisomy 9q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 9q. ORPHA:96101

Distal trisomy 9q is a rare chromosomal anomaly, resulting from the partial trisomy of the long arm of chromosome 9, with a variable phenotype mostly characterized by psychomotor and speech delay, intellectual disability, hypotonia, long narrow habitus, craniofacial dysmorphism (incl. micro/dolichocephaly, facial asymmetry, narrow palpebral fissures, deep-set eyes, strabismus, microphthalmia, abnormally shaped ears, microstomia, micro/retrognathia) and hand and feet anomalies (incl. arachnodactyly, camptodactyly, abnormal implantation of digits). Congenital flexion contractures and limited joint movements have also been observed.