Distal trisomy 20q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 20q. ORPHA:96107

Distal trisomy 20q is a rare chromosomal anomaly syndrome, resulting from the partial trisomy of the long arm of chromosome 20, with high phenotypic variability mostly characterized by neurodevelopmental delay, cardiac malformations (e.g. ventricular septal defect, coarctation of aorta) and facial dysmorphism (incl. large/high forehead, microphthalmia, upslanting palpebral fissures, epicanthus, large, long, low-set ears, anteverted nares, protruding upper lip, cleft lip/palate, micro/retrognathia, dimpled chin). Skeletal (brachydactyly, scoliosis, pectus excavatum) and cerebral anomalies have also been reported.