Distal trisomy 11q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 11q. ORPHA:96103

Distal trisomy 11q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 11, with high phenotypic variability principally characterized by craniofacial dysmorphism (brachycephaly/plagiocephaly, low-set, posteriorly rotated ears, short philtrum, micrognathia) and intellectual disability. Short stature and seizures, as well as cardiac (e.g. atrial septal defect), skeletal (incl. brachy/syndactyly) and genital (e.g. micropenis, cryptorchidism) abnormalities may also be associated. Neurodevelopmental anomalies (pain insensitivity, sensorineural hearing loss, expressive language deficiency) and neuropsychiatric disorders (autistic features, auditory hallucination, self-talking) have also been reported.