Distal monosomy 20q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal monosomy 20q. ORPHA:96152

A rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 20, with a highly variable phenotype typically characterized by global developmental delay with important speech and language deficits, intellectual disability, hypotonia, epilepsy, behavioral anomalies (e.g. autism spectrum disorder behaviors) and hand and feet skeletal malformations. Craniofacial dysmorphism, including microcephaly, high forehead, hypertelorism, broad nasal bridge, bulbous nasal tip, malformed ears, long philtrum, thin upper lip, and microretrognathia, may be occasionally associated.