Distal trisomy 19q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 19q. ORPHA:1717

Distal trisomy 19q is a rare chromosomal anomaly syndrome characterized by low birth weight, developmental delay, intellectual disability, short stature, craniofacial dysmorphism (incl. microcephaly, midface hypoplasia, hypertelorism, flat nasal bridge, ear anomalies, short philtrum, downturned corners of the mouth, micrognathia) and a short neck with redundant skin folds. Additional features may include hypotonia, skeletal anomalies (e.g. clino/camptodactyly), seizures and congenital cardiac, urogenital and gastrointestinal malformations.