Non-distal trisomy 9q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Non-distal trisomy 9q. ORPHA:96112

Non-distal trisomy 9q is a rare chromosomal anomaly syndrome, resulting from the partial trisomy of the long arm of chromosome 9, with a highly variable phenotype principally characterized by developmental delay, short stature, intellectual disability, and craniofacial dysmorphism (e.g. microcephaly, broad forehead, low set ears, epicanthus, prominent nose, and retrognathia). Cardiac, ocular, thyroid and esophagus defects, as well as central nervous system and behavioral/psychiatric abnormalities, have also been reported.