Non-distal trisomy 10q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Non-distal trisomy 10q. ORPHA:1695

Non-distal trisomy 10q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 10, characterized by mild to moderate developmental delay, postnatal growth retardation, central hypotonia, craniofacial dysmorphism (incl. microcephaly, prominent forehead, flat, thick ear helices, deep-set, small eyes, epicanthus, upturned nose, bow-shaped mouth, highly arched palate, micrognathia), ocular anomalies (e.g. iris coloboma, retinal dysplasia, strabismus), long, slender limbs and skeletal and digital anomalies (scoliosis, poly/syndactyly). Additional features reported include cardiac defects (e.g. septal ventricular defect), anal atresia, and cryptorchidism.