Distal trisomy 22q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 22q. ORPHA:96109

Distal trisomy 22q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 22, with variable phenotype principally characterized by varying degrees of intellectual disabilty and developmental delay, pre- and postnatal growth deficiency, hypotonia, and craniofacial dysmorphism (incl. microcephaly, hypertelorism, narrow and upslanted palpebral fissures, epicanthic folds, low-set dysplastic ears, broad and depressed nasal bridge, cleft lip an/or palate, long philtrum, retro/micrognathia). Congenital heart defects, as well as cerebral, skeletal, renal and genital anomalies, have also been reported.