Distal trisomy 13q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 13q. ORPHA:96105

Distal trisomy 13q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 13, with variable phenotype principally characterized by intellectual disability, psychomotor delay, craniofacial dysmorphism (incl. microcephaly, bushy eyebrows, long curled eyelashes, hypotelorism, low-set ears, prominent nasal bridge, long philtrum, high palate, thin upper lip), short neck, polydactyly, and hemangiomas. Cardiac, urogenital and neural tube defects, as well as umbilical and inguinal hernias, seizures and hypotonia, have also been reported.