Distal trisomy 8q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 8q. ORPHA:96100

Distal trisomy 8q is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 8, with a highly variable phenotype, typically characterized by growth and developmental delay, intellectual disability, short stature, craniofacial dysmorphism (microcephaly, prominent forehead, hypertelorism, abnormal palpebral fissures, low-set, large ears, anteverted tip of nose, micro/retrognathia), congenital heart defects and skeletal and limb anomalies. Other reported features include ophthalmologic abnormalities (e.g. megalocornea), cryptorchidism, hypertrichosis, and neurologic manifestations (e.g. hypotonia, hearing loss, and seizures).