Trisomy 1q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Trisomy 1q. ORPHA:261344

Trisomy 1q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 1, with a highly variable phenotype principally characterized by intellectual disability, short stature, craniofacial dysmorphism (incl. macro/microcephaly, prominent forehead, posteriorly rotated, low-set ears, abnormal palpebral fissures, microphthalmia, broad, flat nasal bridge, high-arched palate, micro/retrognathia), cardiac defects and urogenital anomalies. Patients may also present cerebral (e.g. ventriculomegaly) and gastrointestinal malformations, as well as dystonic tremor and recurrent respiratory tract infections.