Distal trisomy 5q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 5q. ORPHA:96097

Distal trisomy 5q is a rare chromosomal anomaly syndrome, resulting from a partial duplication of the long arm of chromosome 5, characterized by short stature, moderate intellectual disability, and craniofacial dysmorphism (microcephaly, flat facies, large, low-set dysplastic ears, down-slanted, almond-shaped palpebral fissures, hypertelorism, epicanthal folds, small nose, long philtrum, small mouth with thin upper lip, and micrognathia). Patients also frequently present speech and cognitive delay, cardiac (ventriculomegaly, ventricular septum defect) and skeletal abnormalities (craniosynostosis, radial agenesis, ulnar hypoplasia, brachydactyly) and genital malformations (hypospadias, cryptorchidism).