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# Distal trisomy 4q

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. [Distal trisomy 4q](#). ORPHA:96096*

Distal trisomy 4q is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 4, with highly variable phenotype typically characterized by psychomotor delay, intellectual disability, craniofacial dysmorphism (microcephaly, low-set, prominent ears, downslanting palpebral fissures, hypertelorism, epicanthic folds, broad, prominent nasal bridge, high arched and cleft palate, micro-/retrognathia), seizures, as well as tooth and digital anomalies (clinodactyly, polydactyly). Cardiac malformations, renal anomalies, cryptorchidism, hypotonia and hearing impairment have also been reported.