# Distal trisomy 2q

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 2q. ORPHA:96094*

Distal trisomy 2q is a rare chromosomal anomaly, resulting from the partial duplication of the long arm of chromosome 2, characterized by moderate psychomotor delay, mild intellectual disability, facial dysmorphism (high hairline, prominent forehead, hypertelorism, upslanting palpebral fissures, large, low-set and/or posteriorly rotated ears, depressed/broad nasal bridge, prominent nasal tip, thin upper lip vermillion), clino-/camptodactyly and normal or increased body measurements. On occasion genital anomalies (hypospadias, cryptorchidism, shawl scrotum) and short stature may be observed.