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# 20q11.2 microduplication syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. 20q11.2 microduplication syndrome. ORPHA:363659*

20q11.2 microduplication syndrome is a rare chromosomal anomaly syndrome, due to partial duplication of the long arm of chromosome 20, characterized by psychomotor and developmental delay, moderate intellectual disability, metopic ridging/trigonocephaly, short hands and/or feet and distinctive facial features (epicanthus, hypoplastic supraorbital ridges, horizontal/downslanting palpebral fissures, small nose with depressed nasal bridge and anteverted nostrils, prominent cheeks, retrognathia and small, thick ears). Growth delay and cryptorchidism are often associated features.