Open Peer Review on Qeios

Non-distal monosomy 10q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Non-distal monosomy 10q. ORPHA:1581

Non-distal monosomy 10q is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 10, with a highly variable phenotype principally characterized by developmental delays (usually of language and speech), variable cognitive impairment and neurobehavioral abnormalities such as autism spectrum disorders and attention deficit disorder. Macrocephaly and mild dysmorphic features may by associated. Overlap with other syndromes, such as Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome and juvenile polyposis syndrome has been reported.