Distal 16p11.2 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal 16p11.2 microdeletion syndrome. ORPHA:261222

Distal 16p11.2 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from the partial deletion of the short arm of chromosome 16 with a highly variable phenotype typically characterized by developmental delay, mild intellectual disability and autism spectrum disorder. Macrocephaly (apparent by 2 years of age), structural brain malformations, epilepsy, vertebral anomalies and obesity are frequently associated.