Distal 17p13.3 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal 17p13.3 microdeletion syndrome. ORPHA:261257

Distal 17p13.3 microdeletion syndrome is a rare partial monosomy of the short arm of chromosome 17 with a variable phenotype characterized by prenatal and postnatal growth retardation, developmental delay, mild intellectual disability, macrocephaly, mild facial dysmorphisms including prominent forehead, hypertelorism, thick upper and/or lower lip vermillion, and structural abnormalities of the brain variably including white matter abnormalities, prominent Virchow-Robin spaces, Chiari I malformation, corpus callosum hypoplasia, but no lissencephaly.