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Proximal 16p11.2 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Proximal 16p11.2 microduplication syndrome. ORPHA:370079

Proximal 16p11.2 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from a partial duplication of the short arm of chromosome 16 characterized by developmental delay and intellectual disability of a highly variable degree, autism spectrum, obsessive-compulsive, attention deficit hyperactivity disorder, speech articulation abnormalities, muscular hypotonia, tremor, hyper- or hyporeflexia, seizures, microcephaly, neuroimaging abnormalities, decreased body mass index and schizophrenia or bipolar disorder later on in life.