19p13.13 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 19p13.13 microdeletion syndrome. ORPHA:357001

19p13.13 microdeletion syndrome is a rare partial autosomal monosomy characterized by global developmental delay, moderate intellectual disability, macrocephaly, overgrowth, hypotonia, and facial dysmorphism (frontal bossing, down-slanting palpebral fissures). Other associated features variably include ataxia, seizures, ventriculomegaly, ocular abnormalities (strabismus, optic nerve hypoplasia) and gastrointestinal problems (abdominal pain, vomiting, constipation).