14q11.2 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 14q11.2 microduplication syndrome. ORPHA:261229

14q11.2 microduplication syndrome is a rare chromosomal anomaly characterized by developmental delay, mild to severe intellectual disability with speech impairment and epilepsy. Additionally, it may include dysmorphic features (such as hypo- or hypertelorism, dysplastic ears, short palpebral fissures), microcephaly or macrocephaly, behavioral abnormalities, stereotyped hand movements, ataxia, hypotonia, cleft palate.