11p15.4 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 11p15.4 microduplication syndrome. ORPHA:300305

11p15.4 microduplication syndrome is a rare partial autosomal trisomy/tetrasomy characterized by obesity, global developmental delay and intellectual disability, facial dysmorphism (synophrys, high-arched eyebrows, large posteriorly rotated ears, upturned nose, long smooth philtrum, overbite and high palate), large hands and limb hypotonia. Additional features include seizures and behavioral abnormalities.