Open Peer Review on Qeios

5p13 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 5p13 microduplication syndrome. ORPHA:329802

5p13 microduplication syndrome is a rare partial autosomal trisomy/tetrasomy characterized by global developmental delay, intellectual disability, autistic behavior, muscular hypotonia, macrocephaly and facial dysmorphism (frontal bossing, short palpebral fissures, low set, dysplastic ears, short or shallow philtrum, high arched or narrow palate, micrognathia). Other associated clinical features include sleep disturbances, seizures, aplasia/hypoplasia of the corpus callosum, skeletal abnormalities (large hands and feet, long fingers and toes, talipes).