
DISEASE:
15q11.2 microdeletion syndrome

NAME:	15q11.2 microdeletion syndrome
DESCRIPTION:	15q11.2 microdeletion syndrome is a rare partial autosomal monosomy with a variable phenotypic expression and reduced penetrance associated with an increased susceptibility to neuropsychiatric or neurodevelopmental disorders including delayed psychomotor development, speech delay, autism spectrum disorder, attention deficit-hyperactivity disorder, obsessive-compulsive disorder, epilepsy or seizures. It may also include mild non-specific dysmorphic features (such as dysplastic ears, broad forehead, hypertelorism), cleft palate, neurological and neuroimaging abnormalities (such as ataxia and muscular hypotonia).
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