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).
ORPHACODE:	261183
SYNONYMS:	15q11.2 BP1-BP2 microdeletion syndrome Del(15)(q11.2) Monosomy 15q11.2
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	NIPA1 NIPA2 TUBG1
CREATED:	17 May 2022 - 02:46

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/4114

RELATED CONTENT

Related Analytes

- NIPA magnesium transporter 1
- NIPA magnesium transporter 2
- tubulin gamma 1

Source URL: http://gentest.healthdata.be/disease/4114