DISEASE:
14q11.2 microduplication syndrome

NAME:	14q11.2 microduplication syndrome
DESCRIPTION:	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.
ORPHACODE:	261229
SYNONYMS:	Dup(14)(q11.2) Trisomy 14q11.2
XREF(S):	Orphanet
ANALYTE(S):	CHD8 SUPT16H FOXG1
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RELATED CONTENT

Related Analytes

- chromodomain helicase DNA binding protein 8
- forkhead box G1
- SPT16 homolog, facilitates chromatin remodeling subunit