DISEASE:
2q23.1 microdeletion syndrome

NAME:	2q23.1 microdeletion syndrome
DESCRIPTION:	The newly described 2q23.1 microdeletion syndrome includes severe intellectual deficit with pronounced speech delay, behavioral abnormalities including hyperactivity and inappropriate laughter, short stature and seizures.
ORPHACODE:	228402
SYNONYMS:	Del(2)(q23.1) Monosomy 2q23.1 Pseudo-Angelman syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MBD5
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- methyl-CpG binding domain protein 5