DISEASE:
2q24 microdeletion syndrome

NAME:	2q24 microdeletion syndrome
DESCRIPTION:	2q24 microdeletion syndrome is a chromosomal anomaly consisting of a partial long arm deletion of chromosome 2 and characterized clinically by a wide range of manifestations (depending on the specific region deleted) which can include seizures, microcephaly, dysmorphic features, cleft palate, eye abnormalities (coloboma, cataract and microphthalmia), growth retardation, failure to thrive, heart defects, limb anomalies, developmental delay and autism.
ORPHACODE:	1617
SYNONYMS:	Del(2)(q24) Monosomy 2q24
XREF(S):	Orphanet MeSH ICD-10
ANALYTE(S):	TBR1
CREATED:	13 May 2019 - 01:02
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