DISEASE:
21q22.11q22.12 microdeletion syndrome

NAME:	21q22.11q22.12 microdeletion syndrome
DESCRIPTION:	A rare, genetic, chromosomal anomaly syndrome resulting from a partial deletion of the long arm of chromosome 21 characterized by pre- and post-natal growth delay, short stature, intellectual disability, developmental delay with severe language impairment, thrombocytopenia, and craniofacial dysmorphism which may include microcephaly, downslanted palpebral ?ssures, low-set ears, broad nose, thin upper vermillion, and downturned corners of the mouth. Brain MRI abnormalities (such as agenesis of the corpus callosum), behavioral problems and seizures may be associated.
ORPHACODE:	261323
SYNONYMS:	Del(21)(q22.11q22.12) Monosomy 21q22.11q22.12
XREF(S):	Orphanet
ANALYTE(S):	KIF15
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- kinesin family member 15