DISEASE:
12q15q21.1 microdeletion syndrome

NAME:	12q15q21.1 microdeletion syndrome
DESCRIPTION:	12q15q21.1 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from a partial deletion of the long arm of chromosome 12, with a highly variable phenotype, typically characterized by developmental delay, learning disability, intra-uterine and postnatal growth retardation, and mild facial dysmorphism that changes with age. Nasal speech and hypothyroidism are also associated.
ORPHACODE:	289513
SYNONYMS:	Del(12)(q15)(q21.1) Deletion 12q15q21.1 Monosomy 12q15q21.1
XREF(S):	Orphanet
ANALYTE(S):	CNOT2
CREATED:	04 Feb 2020 - 15:13
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- CCR4-NOT transcription complex subunit 2