# DISEASE:
# 12q15q21.1 microdeletion syndrome

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|---|---|
| **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)<br>Deletion 12q15q21.1<br>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