# 12q15q21.1 microdeletion syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 12q15q21.1 microdeletion syndrome. ORPHA:289513*

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.