# DISEASE:
# 17q24.2 microdeletion syndrome

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| **NAME:** | 17q24.2 microdeletion syndrome |
| **DESCRIPTION:** | A rare, genetic, multiple congenital anomalies/dysmorphic features-intellectual disability syndrome characterized by developmental and speech delay, intellectual disability, feeding difficulties, failure to thrive, growth retardation, and associated malformations such as abnormality of fingers and toes (i.e. clinodactyly of the 5th finger, 2-3 toe syndactyly), microcephaly, heart defects, and upper airways anomalies. Observed facial dysmorphism includes hypertelorism, small, narrow or downslanting palpebral fissures, ptosis, epicanthus, ear malformations, broad nasal bridge, bulbous/prominent nose, short philtrum, thin lips, retrognathia/micrognathia, arched/cleft palate, and dental anomalies. Additional variable manifestations include hearing and visual impairment, seizures, joint anomalies, obesity, and behavioral/psychiatric disorders. |
| **ORPHACODE:** | 529962 |
| **SYNONYMS:** | Del(17)(q24) |
| **XREF(S):** | Orphanet |
| **ANALYTE(S):** | BPTF<br>PSMD12 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Analytes

- bromodomain PHD finger transcription factor
- proteasome 26S subunit, non-ATPase 12