

## Opis choroby \*

### Definicja

Non-distal trisomy 9q is a rare chromosomal anomaly syndrome, resulting from the partial trisomy of the long arm of chromosome 9, with a highly variable phenotype principally characterized by developmental delay, short stature, intellectual disability, and craniofacial dysmorphism (e.g. microcephaly, broad forehead, low set ears, epicanthus, prominent nose, and retrognathia). Cardiac, ocular, thyroid and esophagus defects, as well as central nervous system and behavioral/psychiatric abnormalities, have also been reported.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Non-distal duplication 9q

Trisomia interstycjalna 9q

Duplikacja interstycjalna 9q

Non-telomeric trisomy 9q

Nietelomerowa duplikacja 9q

#### Kod ORPHA

96112

#### Kod OMIM

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#### Kod ICD10

Q92.3

#### Kod ICD11

LD41.80

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#### \*Źródło

orphanet