

## Opis choroby \*

### Definicja

Non-distal trisomy 13q is a rare chromosomal anomaly disorder, resulting from the partial duplication of the proximal long arm of chromosome 13, with a highly variable phenotype principally characterized by increased polymorphonuclear leucocyte projections and persistence of fetal hemoglobin, as well as growth and developmental delay and craniofacial dysmorphism (incl. microcephaly, depressed nasal bridge, stubby nose, low-set, malformed ears, cleft lip/palate, micrognathia). Strabismus, clinodactyly and undescended testes in males may also be associated.

### Dane

#### Klasyfikacja                      Synonimy

Zespół wad wrodzonych Interstycjalna duplikacja 13q  
Nietelomerowa trisomia 13q  
Non-telomeric trisomy 13q  
Non-distal trisomy 13q

#### Kod ORPHA

1702

#### Kod OMIM

-

#### Kod ICD10

Q92.3

#### Kod ICD11

-

---

#### \*Źródło

orphanet