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
Zespół wad wrodzonych

Synonimy
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