

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	Synonimy
Zespół wad wrodzonych	Non-distal duplication 9q Trisomia interstycjalna 9q Duplikacja interstycjalna 9q Non-telomeric trisomy 9q Nietelomerowa duplikacja 9q

Kod ORPHA 96112	Kod OMIM -	Kod ICD10 Q92.3
Kod ICD11 LD41.80		

*Źródło

orphanet