Opis choroby *

Definicja

Distal 16p11.2 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from the partial deletion of the short arm of chromosome 16 with a highly variable phenotype typically characterized by developmental delay, mild intellectual disability and autism spectrum disorder. Macrocephaly (apparent by 2 years of age), structural brain malformations, epilepsy, vertebral anomalies and obesity are frequently associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal del(16)(p11.2)
Dystalna del(16)(p11.2)
Monosomia dystalna 16p11.2
Distal monosomy 16p11.2

Kod ORPHA
261222

Kod OMIM
613444

Kod ICD10
Q93.5

Kod ICD11
-

---

*Źródło

orphanet