Opis choroby *

Definicja

Monosomy 13q34 is a rare chromosomal anomaly syndrome, resulting from the partial deletion of the long arm of chromosome 13, principally characterized by global developmental delay, mild intellectual disability, obesity and mild craniofacial dysmorphism (microcephaly, wide rectangular forehead, downslanting palpebral fissures, mild ptosis, prominent nose with long nasal bridge and broad tip, small chin). Other variable reported features include congenital heart defects, hand and foot anomalies (e.g. polydactyly) and agenesis of the corpus callosum.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(13)(q34)
Del(13)(q34)
Delecja dystalna 13q34
Delecja subtelomerowa 13q34
Distal deletion 13q34
Subtelomeric deletion 13q34

Kod ORPHA
96168

Kod OMIM
619148

Kod ICD10
Q93.5

Kod ICD11
LD44.D

---

*Źródło

orphanet