

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