Opis choroby *

Definicja

7q11.23 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from the partial duplication of the long arm of chromosome 7 characterized by a highly variable phenotype that typically manifests with mild-moderate intellectual delay (patients could be in the normal range), speech disorders (particularly of expressive language), and distinctive craniofacial features (brachycephaly, broad forehead, straight eyebows, broad nasal tip, short philtrum, thin upper lip and facial asymmetry). Hypotonia, developmental coordination disorders, behavioral problems (such as anxiety, ADHD and oppositional disorders) and various congenital anomalies, such as heart defects, diaphragmatic hernia, renal malformations and cryptorchidism, are frequently presented. Neurological abnormalities (visible on MRI) have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dup(7)(q11.23)
Dup(7)(q11.23)
Trisomia 7q11.23
Trisomy 7q11.23

Kod ORPHA
96121

Kod OMIM
609757

Kod ICD10
Q92.3

Kod ICD11
LD41.60

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*Źródło

orphanet