

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 eyebrows, 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	Synonimy
Zespół wad wrodzonych	Dup(7)(q11.23)
	Dup(7)(q11.23)
	Trisomia 7q11.23
	Trisomy 7q11.23

Kod ORPHA	Kod OMIM	Kod ICD10
96121	609757	Q92.3

Kod ICD11
LD41.60

*Źródło

orphanet