Opis choroby *

Definicja

17p11.2 microduplication syndrome is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 17, typically characterized by hypotonia, poor feeding, failure to thrive, developmental delay (particularly cognitive and language deficits), mild-moderate intellectual deficit, and neuropsychiatric disorders (behavioral problems, anxiety, attention deficit hyperactivity disorder, autistic spectrum disorder, bipolar disorder). Structural cardiovascular anomalies (dilated aortic root, bicommissural aortic valve, atrial/ventricular and septal defects) and sleep disturbance (obstructive and central sleep apnea) are also frequently associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Potocki-Lupski syndrome
Trisomia 17p11.2
Zespół Potockiego i Lupskiego
Trisomy 17p11.2

Kod ORPHA
1713

Kod OMIM
610883

Kod ICD10
Q92.3

Kod ICD11
LD41.G1

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*Źródło

orphanet