Opis choroby *

Definicja

Proximal 16p11.2 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from a partial duplication of the short arm of chromosome 16 characterized by developmental delay and intellectual disability of a highly variable degree, autism spectrum, obsessive-compulsive, attention deficit hyperactivity disorder, speech articulation abnormalities, muscular hypotonia, tremor, hyper- or hyporeflexia, seizures, microcephaly, neuroimaging abnormalities, decreased body mass index and schizophrenia or bipolar disorder later on in life.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Proximal dup(16)(p11.2)
Proksymalna dup(16)(p11.2)
Proksymalna trisomia 16p11.2
Proximal trisomy 16p11.2

Kod ORPHA
370079

Kod OMIM
614671

Kod ICD10
Q92.3

Kod ICD11
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*Źródło

orphanet