Opis choroby *

Definicja

Distal 17p13.3 microdeletion syndrome is a rare partial monosomy of the short arm of chromosome 17 with a variable phenotype characterized by prenatal and postnatal growth retardation, developmental delay, mild intellectual disability, macrocephaly, mild facial dysmorphisms including prominent forehead, hypertelorism, thick upper and/or lower lip vermillion, and structural abnormalities of the brain variably including white matter abnormalities, prominent Virchow-Robin spaces, Chiari I malformation, corpus callosum hypoplasia, but no lissencephaly.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal del(17)(p13.3)
Dystalna del(17)(p13.3)
Monosomia dystalna 17p13.3
Distal monosomy 17p13.3

Kod ORPHA
261257

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet