Opis choroby *

Definicja

The proximal 16p11.2 microdeletion syndrome is a chromosomal anomaly characterized by developmental and language delays, mild intellectual disability, social impairments (autism spectrum disorders), mild variable dysmorphism and predisposition to obesity.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Proximal del(16)(p11.2)
Monosomia proksymalna 16p11.2
Proksymalna del(16)(p11.2)
Proximal monosomy 16p11.2

Kod ORPHA
261197

Kod OMIM
611913

Kod ICD10
Q93.5

Kod ICD11
LD44.G1

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*Źródło

orphanet