Opis choroby *

Definicja

A rare partial autosomal monosomy characterized by language development delay with childhood apraxia of speech, mild intellectual disability, behavourial abnormalities (autistic spectrum disorder, attention deficit hyperactivity disorder, anxiety) and mildly dysmorphic nonspecific features. Additional clinical features may include muscular hypotonia and joint laxity, hernias and microcephaly.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
12p13.33 microdeletion syndrome
Del(12)(p13.33)
Delecja dystalna 12p
Zespół mikrodelecji 12p13.33
Del(12)(p13.33)
Delecja telomerowa 12p
Distal monosomy 12p

Kod ORPHA
280325

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet