

Opis choroby *

Definicja

A rare partial autosomal monosomy characterized by language development delay with childhood apraxia of speech, mild intellectual disability, behavioural 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	Synonimy
Zespół wad wrodzonych 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	Kod OMIM	Kod ICD10
280325	-	Q93.5

Kod ICD11

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*Źródło

orphanet