Opis choroby *

Definicja

Xq12-q13.3 duplication syndrome is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome X, characterized by global developmental delay, autistic behavior, microcephaly and facial dysmorphism (including down-slanting palpebral fissures, depressed nasal bridge, anteverted nares, long philtrum, down-slanting corners of the mouth). Seizures have also been reported in some patients.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Dup(X)(q12-q13.3)
	Dup(X)(q12-q13.3)

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
314389	-	Q99.8

Kod ICD11
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*Źródło

orphanet