

Opis choroby *

Definicja

The newly described 8q12 microduplication syndrome is associated with unusual and characteristic multi-organ clinical features, which include hearing loss, congenital heart defects, intellectual disability, hypotonia in infancy, and Duane anomaly (see this term).

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Dup(8)(q12) Dup(8)(q12) Trisomia 8q12 Trisomy 8q12

Kod ORPHA	Kod OMIM	Kod ICD10
228399	-	Q92.3

Kod ICD11
LD41.70

*Źródło

orphanet