

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

Zespół wad wrodzonych

Synonimy

Dup(8)(q12)

Dup(8)(q12)

Trisomia 8q12

Trisomy 8q12

Kod ORPHA

228399

Kod OMIM

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Kod ICD10

Q92.3

Kod ICD11

LD41.70

*Źródło

orphanet