

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

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

Kod ORPHA

314389

Kod OMIM

-

Kod ICD10

Q99.8

Kod ICD11

-

*Źródło

orphanet