Opis choroby *

Definicja

A rare genetic disorder caused by deletions in the long arm of chromosome 11 (11q) and mainly characterized by craniofacial dysmorphism, congenital heart disease, intellectual disability, Paris Trousseau bleeding disorder, structural kidney defects and immunodeficiency.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
11q terminal deletion syndrome
Del(11)(q23.3)
Del(11)(qter)
Dystalna delecja 11q
Dystalna monosomia 11q
Monosomia 11qter
Telomerowa delecja 11q
Del(11)(q23.3)
Del(11)(qter)
Distal deletion 11q
Distal monosomy 11q
Monosomy 11qter
Telomeric deletion 11q

Kod ORPHA
2308

Kod OMIM
147791

Kod ICD10
Q93.5

Kod ICD11
LD44.B0

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*Źródło

orphanet