Opis choroby *

Definicja

3q13 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from a partial deletion of the long arm of chromosome 3. Phenotype can be highly variable, but it is primarily characterized by significant developmental delay, postnatal growth above the mean, muscular hypotonia and distinctive facial features (such as broad and prominent forehead, hypertelorism, epicantic folds, anti-mongloid slanted eyes, ptosis, short philtrum, protruding lips with a full lower lip, high arched palate). Abnormal hypoplastic male genitalia and skeletal abnormalities are frequently present.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(3)(q13)
Del(3)(q13)
Monosomia 3q13
Monosomy 3q13

Kod ORPHA
1621

Kod OMIM
615433

Kod ICD10
Q93.5

Kod ICD11
LD44.30

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*Źródło

orphanet