Opis choroby *

Definicja

7p22.1 microduplication syndrome is a rare chromosomal anomaly syndrome, resulting from a partial interstitial microduplication of the short arm of chromosome 7, characterized by intellectual disability, psychomotor and speech delays, craniofacial dysmorphism (including macrocephaly, frontal bossing, hypertelorism, abnormally slanted palpebral fissures, anteverted nares, low-set ears, microretrognathia) and cryptorchidia. Cardiac (e.g., patent foramen ovale and atrial septal defect), as well as renal, skeletal and ocular abnormalities may also be associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dup(7)(p22.1)
Dup(7)(p22.1)
Trisomia 7p22.1
Trisomy 7p22.1

Kod ORPHA
314034

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
-

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*Źródło

orphanet