Opis choroby *

Definicja

A rare chromosomal anomaly syndrome, resulting from the partial deletion of the long arm of chromosome 22, outside the DiGeorge critical region. The phenotype is characterized by prematurity, pre- and post-natal growth retardation, developmental delay (particularly speech), mild intellectual disability, variable cardiac defects, and minor skeletal anomalies (such as clinodactyly). Dysmorphic features present in half of the individuals include microcephaly, arched eyebrows, deep set eyes, narrow upslanting palpebral fissures, ear abnormalities (low-set ears, tags and pits), hypoplastic alae nasi, smooth philtrum, down-turned mouth, thin upper lip, retro/micrognatia and pointed chin. For certain very distal deletions including the SMARCB1 gene, there is a risk of developing malignant rhabdoid tumours. Most deletions are de novo .

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal del(22)(q11.2)
Dystalna del(22)(q11.2)
Monosomia dystalna 22q11.2
Distal monosomy 22q11.2

Kod ORPHA
261330

Kod OMIM
611867

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet