

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/micrognathia 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	Synonimy
Zespół wad wrodzonych	Distal del(22)(q11.2) Dystalna del(22)(q11.2) Monosomia dystalna 22q11.2 Distal monosomy 22q11.2

Kod ORPHA	Kod OMIM	Kod ICD10
261330	611867	Q93.5

Kod ICD11

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*Źródło

orphanet