

## Opis choroby \*

### Definicja

20q11.2 microduplication syndrome is a rare chromosomal anomaly syndrome, due to partial duplication of the long arm of chromosome 20, characterized by psychomotor and developmental delay, moderate intellectual disability, metopic ridging/trigonocephaly, short hands and/or feet and distinctive facial features (epicanthus, hypoplastic supraorbital ridges, horizontal/downslanting palpebral fissures, small nose with depressed nasal bridge and anteverted nostrils, prominent cheeks, retrognathia and small, thick ears). Growth delay and cryptorchidism are often associated features.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Dup(20)(q11.2)
	Dup(20)(q11.2)

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
363659	-	Q93.5

**Kod ICD11**  
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### \*Źródło

orphanet