

## Opis choroby \*

### Definicja

Distal monosomy 7q36 is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 7, with a highly variable phenotype typically characterized by holoprosencephaly, growth restriction, developmental delay, facial dysmorphism (facial clefts, prominent forehead, hypertelorism, low-set ears, flat and broad nasal bridge, large mouth), abnormal fingers and palm or sole creases, ocular abnormalities, and other congenital malformations (incl. genital anomalies and caudal deficiency sequence). Cardiopathies have been occasionally reported.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Distal deletion 7q36 Dystalna delecja 7q36 Monosomia 7qter Telomerowa delecja 7q36 Monosomy 7qter Telomeric deletion 7q36

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
1636	-	Q93.5

### Kod ICD11

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### \*Źródło

orphanet