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

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal deletion 7q36
Dystalna delecja 7q36
Monosomia 7qter
Telomerowa delecja 7q36
Monosomy 7qter
Telomeric deletion 7q36

Kod ORPHA
1636

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
-

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*Źródło

orphanet