

Opis choroby *

Definicja

A partial deletion of the long arm of chromosome 17 characterized by hypotonia, growth delay, severe global developmental delay, microcephaly, seizures, congenital heart anomalies, hand and foot anomalies (syndactyly, symphalangism) and dysmorphic facial features, including round face, hypertelorism, upslanting palpebral fissures, and micrognathia. Reported deletions involve regions 17q21-q24.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Dystalna delecja 17q

Monosomia 17qter

Telomerowa delecja 17q

Monosomy 17qter

Telomeric deletion 17q

Distal monosomy 17q

Kod ORPHA

1597

Kod OMIM

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Kod ICD10

Q93.5

Kod ICD11

LD44.H0

*Źródło

orphanet