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
-

Kod ICD10
Q93.5

Kod ICD11
LD44.H0

---

*Źródło

orphanet