Opis choroby *

Definicja

An autosomal anomaly characterized by variable clinical features, depending on the size and precise location of deleted chromosome segments. Most patients present with developmental delay, intellectual disability, growth retardation, microcephaly, clinodactyly, and dysmorphic features. Congenital heart disease and genitourinary anomalies were reported in some cases.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ring 10
Ring chromosome 10

Kod ORPHA
1438

Kod OMIM
-

Kod ICD10
Q93.2

Kod ICD11
-

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*Źródło

orphanet