Opis choroby *

Definicja

Ring chromosome 19 syndrome is a rare chromosomal anomaly syndrome with a highly variable phenotype that may range from normal to patients with profound intellectual disability, developmental delay, learning disability (esp. speech) and mild dysmorphism (incl. micro/macrocephaly, prominent forehead, low-set and posteriorly rotated ears, hypertelorism, high nasal bridge, prominent philtrum, retro/micrognathia). Mild hypotonia and autistic-like mannerisms (e.g. hand opening and closing, head banging) may also be associated. Other anomalies, such as cutis laxa, hearing loss, syndactyly, digital hypoplasia, and talipes equinovarus, have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ring 19
Ring chromosome 19

Kod ORPHA
1443

Kod OMIM
-

Kod ICD10
Q93.2

Kod ICD11
-

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*Źródło

orphanet