Opis choroby *

Definicja

Ring chromosome 3 syndrome is a rare chromosomal anomaly syndrome with a highly variable phenotype principally characterized by pre- and postnatal growth retardation, short stature, developmental delay, mild to severe intellectual disability, microcephaly and mild dysmorphic features (incl. triangular face, dysplastic ears, upslanting palpebral fissures, epicanthic folds, broad nasal bridge, full nasal tip, long philtrum, downturned corners of the mouth, and micro/retrognathia). Additional manifestations reported include hypotonia, mild articular limitation, hearing loss, digital anomalies (i.e. clinodacytyly, brachydactyly), café-au-lait patches and hypospadias.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ring 3
Ring chromosome 3
Ring 3
Ring chromosome 3

Kod ORPHA
96172

Kod OMIM
-

Kod ICD10
Q93.2

Kod ICD11
-

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*Źródło

orphanet