

Opis choroby *

Definicja

Ring chromosome 2 syndrome is a rare chromosomal anomaly syndrome with highly variable phenotype principally characterized by intrauterine growth retardation, failure to thrive, developmental delay, hypotonia, mild dysmorphic features (incl. microcephaly, short forehead, upslanting palpebral fissures, hypertelorism, epicanthal folds, wide nasal bridge, broad nasal tip, long philtrum, thin upper lip, micrognathia, short neck), skeletal anomalies (e.g. kyphosis, brachydactyly, clinodactyly, talipes equinovarus) and dermatological features (i.e. café-au-lait spots). Patients may also present ventriculoseptal defects and genital abnormalities (e.g. genital hypoplasia, phimosis, cryptorchidism).

Dane

Klasyfikacja

Zespół wad wrodzonych Ring 2

Synonimy

Ring chromosome 2

Ring 2

Ring chromosome 2

Kod ORPHA

96171

Kod OMIM

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Kod ICD10

Q93.2

Kod ICD11

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*Źródło

orphanet