

Opis choroby *

Definicja

Ring chromosome 5 syndrome is a rare chromosomal anomaly syndrome, with high phenotypic variability, principally characterized by a neonatal mewing cry, severe developmental delay and intellectual disability, short stature, hypotonia, dysmorphic features (incl. microcephaly, facial asymmetry, hypertelorism, epicanthal folds, abnormal ears, micro/retrognathia), congenital cardiac anomalies (such as atrial and ventricular septal defect, tricuspid insufficiency, hypoplastic aorta) and skeletal abnormalities (e.g. hypoplastic thumbs, anomalous ulna/radius, dysplastic metacarpals and phalanges).

Dane

Klasyfikacja

Zespół wad wrodzonych Ring 5

Synonimy

Ring chromosome 5

Kod ORPHA

251043

Kod OMIM

-

Kod ICD10

Q93.2

Kod ICD11

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*Źródło

orphanet