Opis choroby *

Definicja

Mosaic Trisomy 4 is a rare autosomal anomaly, due to the presence of an extra copy of chromosome 4 in a fraction of all cells, with a variable phenotype characterized by intrauterine growth retardation, low birth weight/length/OFC, mild intellectual deficit, congenital heart defects, hypertrophic cardiomyopathy, dysmorphic features (asymmetry of the face, eyebrow anomalies, low-set, posteriorally rotated, dysplastic ears, micro-/retrognathia), characteristic thumb abnormalities (aplasia, hypoplasia) and skin abnormalities (hypo/hyperpigmentation). Delayed puberty may be associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 4
Trisomy 4 mosaicism
Mosaic trisomy chromosome 4
Trisomy 4 mosaicism

Kod ORPHA
96059

Kod OMIM
-

Kod ICD10
Q92.1

Kod ICD11
LD40.Y

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*Źródło

orphanet