

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, posteriorly rotated, dysplastic ears, micro-/retrognathia), characteristic thumb abnormalities (aplasia, hypoplasia) and skin abnormalities (hypo/hyperpigmentation). Delayed puberty may be associated.

Dane

| Klasyfikacja | Synonimy |
|-----------------------|--|
| Zespół wad wrodzonych | Mosaic trisomy chromosome 4 Trisomy 4 mosaicism Mosaic trisomy chromosome 4 Trisomy 4 mosaicism |

| Kod ORPHA | Kod OMIM | Kod ICD10 |
|----------------------------|----------|-----------|
| 96059 | - | Q92.1 |
| Kod ICD11 LD40.Y | | |

*Źródło

orphanet