

Opis choroby *

Definicja

Mosaic trisomy 14 is a rare chromosomal anomaly disorder, with a highly variable phenotype, principally characterized by growth and developmental delay, intellectual disability, body asymmetry/hypotonia, congenital heart defects, genitourinary abnormalities (cryptorchidism, micropenis, large clitoris, labial swelling), and abnormal skin hyperpigmentation. Patients usually present with craniofacial dysmorphism such as microcephaly, abnormal palpebral fissure, hypertelorism, ear abnormalities, broad nose, low-set ears, micro/retro-gnathia, and cleft or highly arched palate.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Mosaic trisomy chromosome 14

Trisomy 14 mosaicism

Kod ORPHA

1703

Kod OMIM

-

Kod ICD10

Q92.1

Kod ICD11

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*Źródło

orphanet