

Mozaikowa trisomia 14

Kod Orpha: 1703 Kod OMIM:

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

Zespół wad wrodzonych

Synonimy

Mosaic trisomy chromosome 14
Trisomy 14 mosaicism

Kod ORPHA

1703

Kod OMIM

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Kod ICD10

Q92.1

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl