Trisomia mozaikowa 7
Kod Orpha: 1747 Kod OMIM:

Opis choroby *

Definicja
Mosaic trisomy 7 is a rare chromosomal anomaly syndrome, with a highly variable phenotype, mostly characterized by blaschkolinear skin pigmentary dysplasia, body asymmetry, enamel dysplasia, and developmental and growth delay. Intellectual disability, facial dysmorphism (e.g. frontal bossing, abnormal palpebral fissures, strabismus, abnormally shaped ears, and micrognathia), and genital anomalies (e.g. undescended testes) have also been observed. It has been reported to be associated with maternal uniparental disomy of chromosome 7, resulting in a Silver-Russell syndrome phenotype. Cases with no associated malformations have also been reported.
Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 7
Trisomy 7 mosaicism

Kod ORPHA
1747

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.

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