Mozaikowa trisomia 1
Kod Orpha: 1692 Kod OMIM:

Opis choroby *

Definicja
A rare autosomal trisomy, characterized by reduced fetal movements and intrauterine growth retardation, low birth weight, and multiple congenital anomalies. The latter include, amongst others, facial dysmorphism (like hypertelorism, cleft lip/palate, micrognathia, low hairline, and small, low-set, and posteriorly rotated ears), head circumference below average, deformities of the hands (camptodactyly) and feet, marked hypertrichosis, and anomalies of the brain, heart, and lungs. Lethality appears to depend on the degree of mosaicism.
Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 1
Trisomy 1 mosaicism

Kod ORPHA
1692

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.