

# Mozaikowa trisomia 17

Kod Orpha: 1711 Kod OMIM:

## Opis choroby \*

### Definicja

Mosaic trisomy 17 is a rare chromosomal anomaly syndrome, with a highly variable clinical presentation, mostly characterized by growth delay, intellectual disability, body asymmetry with leg length differentiation, scoliosis, and congenital heart anomalies (e.g. ventricular septal defect). Prenatal ultrasound findings include intrauterine growth retardation, nuchal thickening brain anomalies (e.g. cerebellar hypoplasia), pleural effusion and single umbilical artery. Patients with no associated malformations have also been reported.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Mosaic trisomy chromosome 17  
Trisomy 17 mosaicism

#### Kod ORPHA

1711

#### 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.