

## **Opis choroby \***

### Definicja

Mosaic trisomy 15 is a rare chromosomal anomaly syndrome principally characterized by intrauterine growth restriction, congenital cardiac anomalies (incl. ventricular and atrial septal defects, patent ductus arteriosus) and craniofacial dysmorphism (incl. hypertelorism, downslanting palpebral fissures, wide nasal bridge). Patients also present brain (e.g. hypoplastic cerebellum, ventricular asymmetry), renal (e.g. small dysplastic kidneys), and/or genital (undescended testis, small penis, hypoplastic labia majora) anomalies. Digital and skin pigmentation abnormalities have also been reported.

### Dane

#### **Klasyfikacja**

#### Synonimy

Zespół wad wrodzonych Mosaic trisomy chromosome 15

Trisomy 15 mosaicism

#### **Kod ORPHA**

1706

#### **Kod OMIM**

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#### **Kod ICD10**

Q92.1

#### **Kod ICD11**

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\*[Źródło](#)

orphanet