

# Mozaikowa trisomia 3

Kod Orpha: 100071 Kod OMIM:

## Opis choroby \*

### Definicja

Mosaic trisomy 3 is a rare chromosomal anomaly syndrome with high phenotypic variability ranging from a mild phenotype presenting joint pain and laxity, mild facial dysmorphism (e.g. long facies, prominent eyes, dysplastic ears, downturned corners of the mouth, micrognathia) and no developmental delays to more severe phenotypes including short stature, intellectual disability, severe developmental delays, additional craniofacial dysmorphic features (e.g. brachycephaly, high forehead, flat midface, short neck) and hearing impairment, as well as skeletal (e.g. pectus excavatum, scoliosis), ocular (e.g. coloboma) and cardiac abnormalities.

Dane

### Klasyfikacja

Zespół wad wrodzonych

### Synonimy

Mosaic trisomy chromosome 3  
Trisomy 3 mosaicism  
Mosaic trisomy chromosome 3  
Trisomy 3 mosaicism

### Kod ORPHA

100071

### Kod OMIM

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### Kod ICD10

Q92.1

### Kod ICD11

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## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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