

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	Synonimy
Zespół wad wrodzonych	Mosaic trisomy chromosome 3 Trisomy 3 mosaicism Mosaic trisomy chromosome 3 Trisomy 3 mosaicism

Kod ORPHA	Kod OMIM	Kod ICD10
100071	-	Q92.1

Kod ICD11

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*Źródło

orphanet