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
-

Kod ICD10
Q92.1

Kod ICD11
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*Źródło

orphanet