

Opis choroby *

Definicja

Mosaic trisomy 20 is a rare chromosomal anomaly syndrome with a highly variable phenotype ranging from normal (in the majority of cases) to a mild, subtle phenotype principally characterized by spinal abnormalities (i.e. stenosis, vertebral fusion, and kyphosis), hypotonia, lifelong constipation, sloped shoulders, skin pigmentation abnormalities (i.e. linear and whorled nevoid hypermelanosis) and significant learning disabilities despite normal intelligence. More severe phenotypes, with patients presenting psychomotor and speech delay, mild facial dysmorphism, cardiac (i.e. ventricular septal defect, dysplastic tricuspid mitral valve) and renal anomalies (e.g. horseshoe kidneys), have also been reported.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Mosaic trisomy chromosome 20 Trisomy 20 mosaicism

Kod ORPHA	Kod OMIM	Kod ICD10
1724	-	Q92.1

Kod ICD11

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*Źródło

orphanet