Trisomia mozaikowa 20
Kod Orpha: 1724 Kod OMIM:

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
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 20
Trisomy 20 mosaicism

Kod ORPHA
1724

Kod OMIM
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Kod ICD10
Q92.1

Kod ICD11
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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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