Opis choroby *

Definicja

Mosaic trisomy 5 is a rare chromosomal anomaly syndrome with a variable phenotype ranging from clinically normal to patients presenting intrauterine growth retardation, congenital heart anomalies (mainly ventricular septal defect), multiple dysmorphic features (e.g. hypertelorism, prominent nasal bridge) and other congenital anomalies (incl. eventration of diaphragm, agenesis of corpus callosum, cloverleaf skull, clinodactyly, anteriorly placed anus). Psychomotor development may be normal in spite of low growth parameters being associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 5
Trisomy 5 mosaicism
Mosaic trisomy chromosome 5
Trisomy 5 mosaicism

Kod ORPHA
96060

Kod OMIM
-

Kod ICD10
Q92.1

Kod ICD11
LD40.Y

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*Źródło

orphanet