

Opis choroby *

Definicja

Maternal uniparental disomy of chromosome 20 (UPD 20) is a very rare chromosomal anomaly in which both copies of chromosome 20 are inherited from the mother. The main feature described is prenatal and postnatal growth retardation. Microcephaly, minor dysmorphic features and psychomotor developmental delay have been occasionally reported. Maternal UPD20 is most often ascertained by a mosaic trisomy 20 pregnancy.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy
Maternal UPD(20)
UPD(20)mat
Matczyna UPD(20)
UPD(20)mat

Kod ORPHA

96186

Kod OMIM

617352

Kod ICD10

Q99.8

Kod ICD11

LD45.0

*Źródło

orphanet