

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	Synonimy
Zespół wad wrodzonych	Maternal UPD(20) UPD(20)mat Matczyna UPD(20) UPD(20)mat

Kod ORPHA	Kod OMIM	Kod ICD10
96186	617352	Q99.8

Kod ICD11
LD45.0

*Źródło

orphanet