

Opis choroby *

Definicja

Paternal uniparental disomy of chromosome 20 is a very rare chromosomal anomaly in which both copies of chromosome 20 are inherited from the father. The main features described are high birth weight and/or early-onset obesity, relative macrocephaly, and tall stature. Most patients were ascertained during sporadic pseudohypoparathyroidism type 1b (see this term) testing and have UPD involving variable segments of the long arm of chromosome 20.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Paternal UPD(20) Ojcowska UPD20 UPD(20)pat UPD(20)pat

Kod ORPHA	Kod OMIM	Kod ICD10
96194	-	Q99.8

Kod ICD11
LD45.1

[*Źródło](#)

orphanet