

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

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

Synonimy

Kod ORPHA

96194

Kod OMIM

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Kod ICD10

Q99.8

Kod ICD11

LD45.1

[*Źródło](#)

orphanet