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Paternal uniparental disomy of chromosome 20

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Paternal uniparental disomy of chromosome 20. ORPHA:96194

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.