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## CASE REPORT

# A Fetus with Maternal Uniparental Disomy on Chromosome 20: Case Report with Genetic Analysis and Prenatal Diagnosis

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**SUMMARY**

***Background:*** **A fetus with increased copy number of chromosome 20 was identified by NIPT. Here we utilize several genetic tests and analyses to illuminate the etiology of such aneuploidy.**
***Methods:*** **Amniotic fluid cells were extracted from pregnant woman and sent for karyotype and chromosomal microarray analysis (CMA). Trio pedigree analysis was conducted with Chromosome Analysis Suite and uniparental disomy (UPD)-tool software.**
***Results:*** **CMA identified consistent results, which were 2 regions of homozygosity: arr[GRCh37]20p12.2q11.1 (11265096_26266313)hmz and arr[GRCh37]20q11.21q13.2(29510306_54430467)hmz. The trio pedigree analysis discovered that the fetal chromosome 20 was the entire maternal UPD mosaic with isodisomy and heterodisomy.**
***Conclusions:*** **When a large segment of chromosome is homozygous, appropriate genetic tests are required to find the potential mechanisms for UPD formation.**
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## Supplementary Data

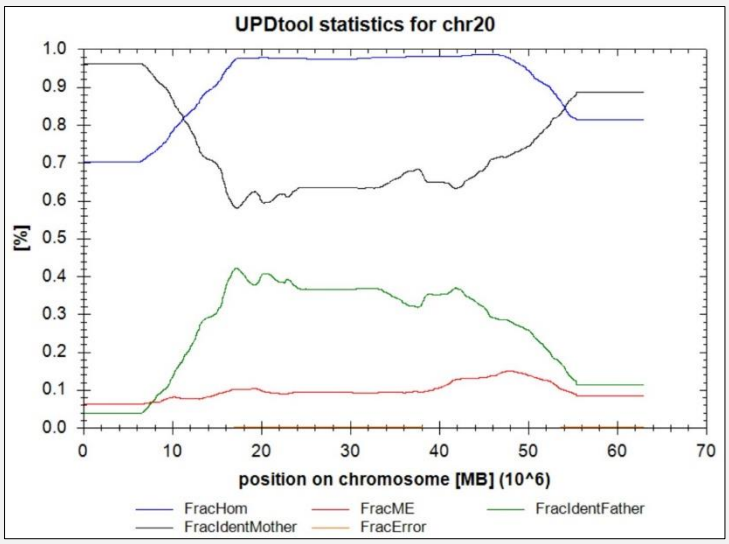


**Figure S1. Uniparental disomy analysis of the pedigree.**

**The homozygous fragment is represented by FracHom (blue line); the Mendelian error fragment is represented by FracME (red line); the fragment identical to the father's allele is represented by FracIdentFather (green line); the fragment identical to the mother's allele is represented by FracIdentMother (black line); fragments that cannot be interpreted by UPD are indicated by FracError (yellow line).**

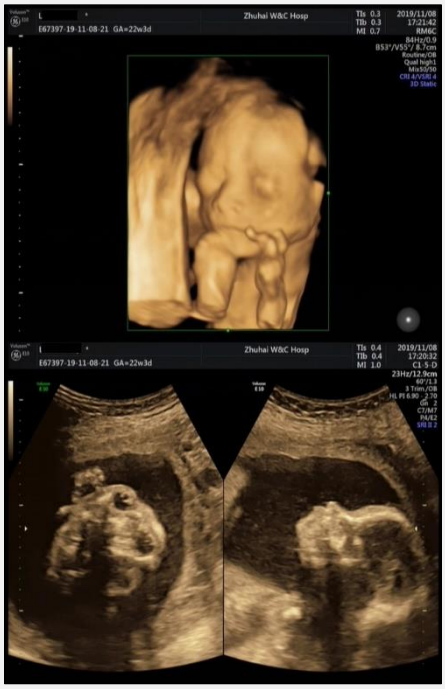

**Figure S2. Ultrasonography at the 22 weeks and 3 days of pregnancy.**

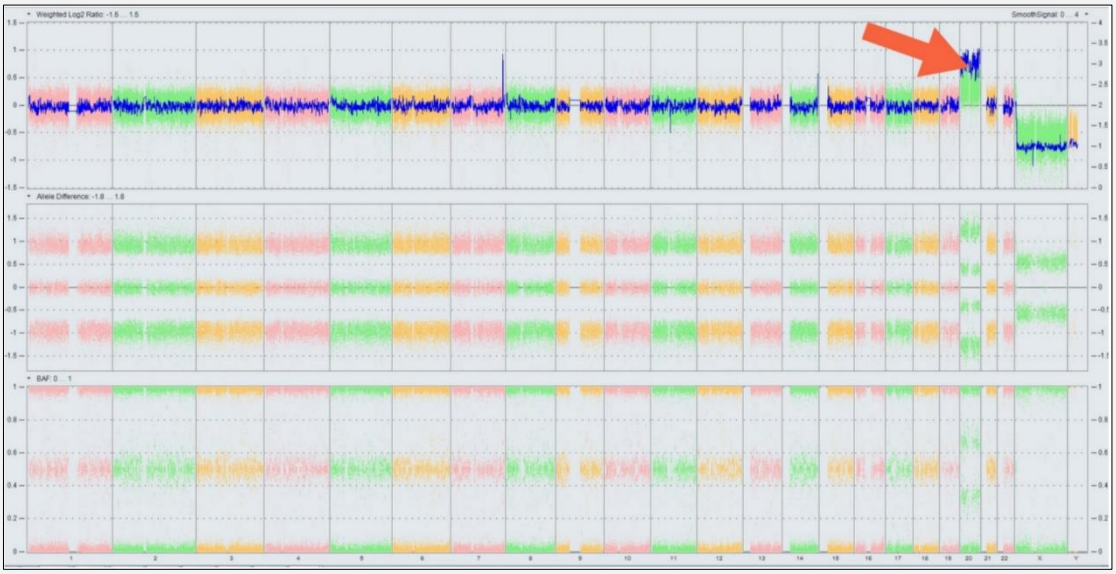

**Figure S3. CMA results of placental tissue revealed trisomy 20.**

**CMA - chromosomal microarray analysis.**