Adding another "hatched pink" chromosome

To the Editor:

In the "American College of Medical Genetics Statement on Diagnostic Testing for Uniparental Disomy," Shaffer et al.[1] show a very clear and practical scheme (Fig. 3) of a human karyotype in which the chromosomes are marked differently if uniparental disomy (UPD) for any of them has been reported in the literature; their effect on the phenotype is also described. In the scheme, those chromosomes in which UPD was never described—for example, chromosome 4—are represented in solid black and those with UPD observed in phenotypically normal individuals are represented as hatched pink.

We wish to point out a case of maternal UPD 4, observed by us in 2000 and published in January 2001.[2] In brief, the case was of a woman in whom chorionic villi sampling (CVS) was performed; a full trisomy 4 was found in the short-term culture, and a normal karyotype was found in both long-term and amniotic fluid cell cultures.

An ultrasound scan in the 29th week showed a fetus with severe intrauterine growth restriction (<5th percentile) and oligohydramnios, who died in utero a few days later. Postmortem examination did not show any malformation. Samples from several sites in the placenta, fetal tissues, and blood from the parents were sent to the Department of Medical Genetics, University of British Columbia, Vancouver, BC, Canada; maternal UPD 4 was found typing nine microsatellites. This was the first case of UPD 4 reported in the literature. It emphasizes the important contribution of CVS studies to the knowledge of UPD in humans, the consequences of UPD, the relevance of cytogenetics studies of the placenta, and the importance of looking for UPD in cases of intrauterine growth restriction of unknown origin.

With this letter we do not intend to lessen the value of the ACMG statement on UPD. Because the two reports were published at around the same time, the authors could not have known about ours. On the contrary, we think that the statement should be updated as new cases of UPD appear in the literature.

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References

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2. Kuchinka BD, Barrett IJ, Moya G, Sánchez JM, Langlois S, Yong SL, Kalousek DK, Robinson WP. Two cases of confined placental mosaicism for chromosome 4, including one with maternal uniparental disomy. *Prenat Diagn* 2001;21:36–39.