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Research Letter

Mosaic isochromosome 20q detected at amniocentesis: A likely cell culture artifact

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A 37-year-old, gravida 3, para 2, woman underwent amniocentesis at 17 weeks of gestation because of advanced maternal age. In five of 26 separated colonies of the cultured amniocytes, an abnormal karyotype of 46,XY,i(20)(q10) (Fig. 1) was noted, while the other 21 colonies had a karyotype of 46,XY. Thus, the conventional cytogenetic result of the cultured amniocytes was 46,XY,i(20)(q10)[5]/46,XY[21]. The parental karyotypes were normal. The prenatal ultrasound findings were unremarkable.

The patient underwent repeated amniocentesis at 22 weeks of gestation. The aspired amniotic fluid was sent to a different laboratory for genetic analysis. Interphase fluorescence *in situ* hybridization (FISH) analysis of the uncultured amniocytes using a 20q13.33-specific probe (RP11-266K16) in the red spectrum and a 20p13-specific probe (RP11-530N10) in the green spectrum revealed two red signals and two green signals in all 50 uncultured amniocytes, indicating no evidence of isochromosome 20q (Fig. 2). Cytogenetic analysis of the cultured amniocytes in the repeated amniocentesis revealed a karyotype of 46,XY in 23 colonies without the mosaic isochromosome 20q finding from previous amniotic fluid culture. The woman decided to continue the pregnancy.

At 38 weeks of gestation, a 3290-g healthy male baby was delivered uneventfully. Cytogenetic analysis of the cord blood revealed a karyotype of 46,XY (50 cells). Interphase FISH analysis of the uncultured urinary cells using the 20q13.33-specific probe RP11-266K16 and the 20p13-specific probe RP11-530N10 showed two red signals and two green signals in all 100 urinary cells, respectively, indicating no evidence of isochromosome 20q (Fig. 3). At routine pediatric follow-up at 6 months of age, the neonate exhibited normal growth and psychomotor development.

Although several reports have described the association of phenotypic abnormalities with mosaic isochromosome 20q detected at amniocentesis [1–4], most reported cases with prenatally-detected mosaicism for isochromosome 20q have presented with normal outcomes [3,5–7]. Robinson et al [6] suggested that the cell line of isochromosome 20q arises through a post-zygotic error and persists only in a few specific cell types. Chen et al [7] observed cytogenetic discrepancy between uncultured and cultured amniocytes in mosaic isochromosome 20q detected at amniocentesis. The present case provides evidence that mosaic isochromosome 20q detected at amniocentesis may be a cell culture artifact. Applying interphase FISH on uncultured amniocytes under such a circumstance allows for a rapid differential diagnosis of

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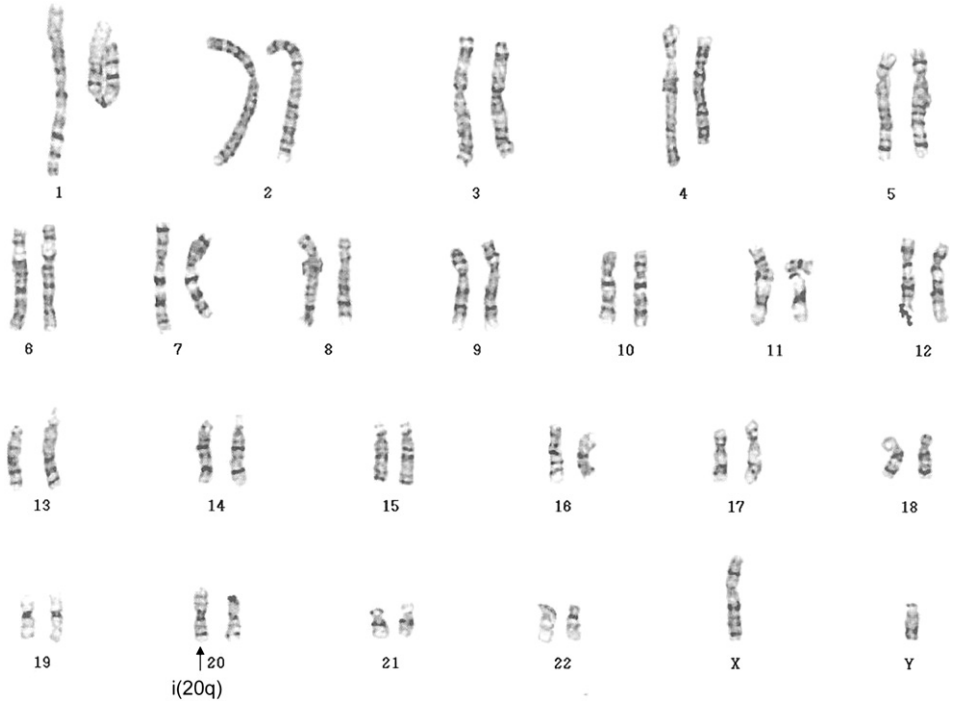

Fig. 1. A karyotype of 46,XY,i(20)(q10).

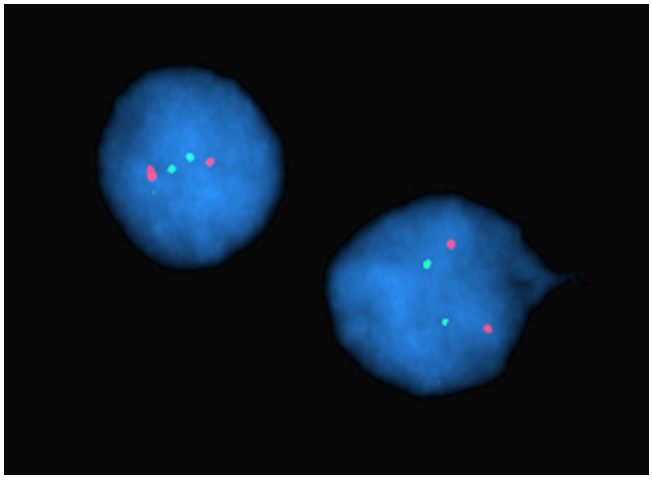

Fig. 2. Interphase fluorescence *in situ* hybridization analysis of uncultured amniocytes using a 20q-specific probe RP11-266K16 (20q13.33) in the red spectrum and a 20p-specific probe RP11-530N10 (20p13) in the green spectrum shows two red signals and two green signals in the uncultured amniocytes, indicating disomy 20 in the cells.

a cell culture artifact while the process of amniocyte cultures is still going on.

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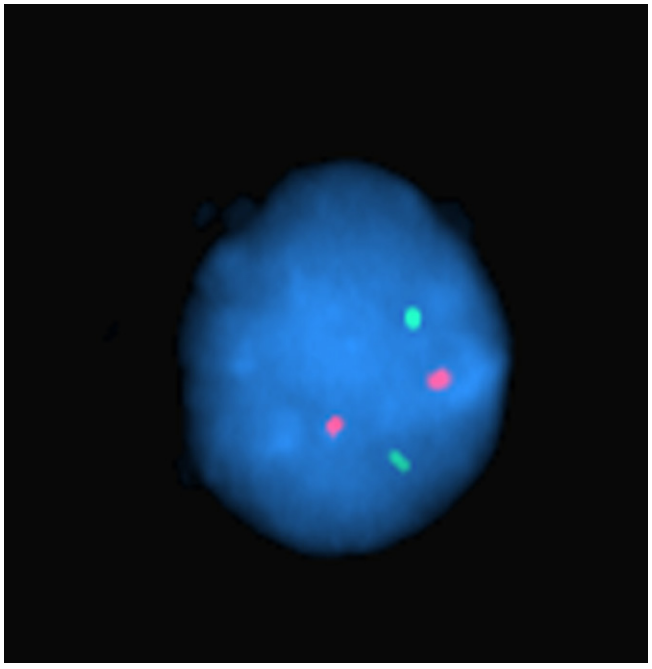

Fig. 3. Interphase fluorescence *in situ* hybridization analysis of uncultured urinary cells using a 20q-specific probe RP11-266K16 (20q13.33) in the red spectrum and a 20p-specific probe RP11-530N10 (20p13) in the green spectrum shows two red signals and two green signals in the uncultured urinary cell, respectively, indicating disomy 20 in the cell.

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