Author's response to reviews

Title: The identification of a spontaneous 47,XX,+21/46,XY chimeric fetus with male genitalia

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Author's response to reviews: see over
Dear Editors:

I am pleased to resubmit the revised manuscript titled “The identification of a 47,XX,+21/46,XY chimeric fetus with male genitalia” by Lee et al. for publication as a “Case Report” in BMC Medical Genetics. The contents of this version have been revised according to reviewers’ comments and a point-by-point responding letter is uploaded to the online submission system. All co-authors have read and agreed to the revised manuscript and there is no conflict of interest in this work. We certify that the resubmission is not under review by any other journal.

In this revised manuscript, we report a chimera case with a karyotype of 47,XX,+21/46,XY that was aborted by induction at the 21st week of gestation after the parents received genetic counseling. The aborted fetus was first examined by autopsy followed by the karyotype analyses of cells that had been cultured from various tissues, including the cerebral cortex, kidney, skin, and placenta. The autopsy demonstrated that all of the examined tissues were normally developed, and the genitalia appeared to be male at this stage. Karyotyping revealed the same chimeric chromosomal composition as that detected by amniocentesis but with a different ratio of (47,XX,+21) to (46,XY) cells. Finally, a short tandem repeat (STR) analysis of the extracted genomic DNA was performed to trace the parental origins for 19 selected markers. Results indicated that the additional chromosome 21 in the 47,XX,+21 cell lineage was of paternal origin, with four markers revealing a two paternal and single maternal contributions. These results indicate that this case represents a chimera formed from dispermice fertilization of a pathenogenic ovum with one (X) sperm and another (Y,+21) sperm.

This is the fifth reported chimera case with trisomy 21 since 1962 and is the only case formed via dispermice fertilization of a pathenogenic ovum. We hope that the editorial board and the reviewers will agree with our replies and the quality of the revised version.

We look forward to the final decision.

Sincerely yours,

Ingrid Y.-C. Liu on behalf of the authors

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