Author's response to reviews

Title: A natural-fertilized 47,XX,+21/46,XY parthenogenetic chimeric fetus with male genitalia

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Version: 2 Date: 19 April 2012

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Dear Editors:

I am pleased to resubmit the manuscript titled “A natural-fertilized 47,XX,+21/46,XY parthenogenetic chimeric fetus with male genitalia” by Lee et al. for publication as a “Case Report” in BMC Medical Genetics. All co-authors have read and agreed to the contents of the manuscript and there is no conflict of interest in this work. We certify that the submission is not under review by any other journal.

In the resubmitted version, we have 1) confirmed that the Institutional Review Board of the Tzu-Chi general hospital approved this study (approval number: IRB098-82); and 2) moved the “Ethics statement” to be the first paragraph of the METHODS section.

In this manuscript, we report a sex-chromosome chimera 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 abortus 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, including the male genitalia. 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 in all examined tissues. Finally, a short-tandem-repeat (STR) analysis of the extracted genomic DNA was performed to trace the parental origins for 19 selected markers. The results indicated that the additional chromosome 21 in the 47,XX,+21 cell lineage was of paternal origin, with some markers revealing one maternal and two paternal contributions. These results indicate that this case represents a parthenogenetic chimera.

This is the fifth reported sex-chromosome chimera case with trisomy 21 since 1962 and is the only parthenogenetic case that the paternal origin of the extra chromosome 21 was identified. We hope that the editorial board and the reviewers will find this report interesting.

Sincerely yours,

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

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