LETTER TO THE EDITOR / EDİTÖRE MEKTUP
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COMMENT ON "A PARTIAL TRISOMY 9 CASE WITH DICENTRIC CHROMOSOME DUE TO THE ADJACENT-2 SEGREGATION OF MATERNAL RECIPROCAL TRANSLOCATION"

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Dear Editor,

We have read the article entitled "A partial trisomy 9 case with dicentric chromosome due to the adjacent-2 segregation of maternal reciprocal translocation" by Urtekin et al. published in the Journal of Istanbul Faculty of Medicine (1). This paper is exceptional because the offspring of adjacent-2 segregation being born alive is a rarity. Although the authors have been careful with the details of each aspect, we would like to comment on certain possible theoretical imbalances, especially those caused by tertiary segregations. The authors state that adjacent-2 segregation is one of the few cases in which the infant is born alive with dysmorphic features as it results in trisomy/monosomy of chromosome 22 with partial deletion/duplication of chromosome 9. Moreover, in 3:1 segregation, there are two possibilities of disomy 22 with duplication and deletion of chromosome 9 derived from the tertiary monosomy/trisomy segregation pattern and that most gestational products will be monosomic or double trisomic for chromosomes 9 and 22 (interchange monosomy/trisomy products).

As the authors have correctly indicated, in this translocation, adjacent-1 segregation ironically results in gametes with greater imbalance than adjacent-2 segregation. Nonetheless, in tertiary 3:1 segregations, the only possibility for disomy of chromosome 22 is caused by gametes with 24 chromosomes retaining der(9). Such gametes will additionally have a disomy of 9q22.31-pter, whereas those with 24 chromosomes and der (22) will have a disomy of 9q22.31-qter only. The gametes with 22 chromosomes retaining der(22), namely -9, will have virtually complete nullisomy for chromosome 22 and nullisomy for 9q22.31-pter, and those retaining der(9), that is -22, will be nullisomic for 9q22.31-qter only. In such cases, the hypothetical offspring of these gametes will not conform to the expected imbalance in tertiary 3:1 segregation. However, the interchange aneuploidies would be the same as those for any other reciprocal translocation (trisomy or monosomy for each chromosome) and not monosomy or double trisomy, as suggested by the authors.

Furthermore, although it is appropriate to inform the family about reproductive options to avoid the recurrence of unbalanced offspring, the fact that this couple may also have phenotypically normal offspring, either with a normal karyotype or chromosomally balanced (like the mother) owing to an alternate segregation, should be noted. Finally, if the maternal grandparents are alive, a karyotype can be requested, and if one of them is a carrier of the translocation, karyotyping of other members can be recommended if they have it.

We believe that these comments do not discredit the study; rather, they complement it.

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