LETTER TO THE EDITOR / EDİTÖRE MEKTUP
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REPLY TO LETTER TO THE EDITOR BY MEZA-ESPINOZA AND COLLEAGUES

MEZA-ESPINOZA VE ARKADAŞLARININ EDİTÖRE MEKTUBUNA CEVAP

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Dear Editor,

In this journal, we reported a rare live birth case of partial trisomy 9 resulting from a maternal translocation between chromosomes (Chr) 9 and 22 (1). The affected infant had two normal Chr9 plus a derived/dicentric Chr9 containing a complete copy of 9pter to 9q22.31 (carrying the centromere regions of both Chr9 and Chr22). Meza-Espinoza and colleagues wrote a letter to the editor about this case and the related translocation-derived gametes (2). We appreciate the contributions of the authors.

All possible gametes are shown in Table 1, and there are two options for disomy of 22 products derived from tertiary 3:1 segregation. However, these gametes are also trisomic or monosomic on the long arm of Chr9 (q22.31 to 9qter). In the case of tertiary trisomy 3:1 segregation, when normal Chr9, normal Chr22 and der(22) are transmitted to the next generation, the gamete will be disomic for Chr22 and partially trisomic for long arm of Chr9 (9q22.31 to 9qter). Trisomy of the short arm of Chr9 is a rare condition but is compatible with life, as in this case. In contrast, the trisomy of the long arm of Chr9 (the product of a tertiary 3:1 segregation) is almost always incompatible with life. In tertiary monosomy 3:1 segregation, when only der(9) is inherited, the offspring will be disomic for Chr22 but will also have partial monosomy from 9q22.31 to 9qter and will have no chance of a live birth. Two disomy 22 products derived from the segregation of interchange trisomy and interchange monosomy also exist, but the gametes will also be trisomy and monosomy for Chr9. Additionally, the second option for the adjacent 2 segregation pattern leads to partial monosomy of chromosome 9 (9pter to 9q22.31, our case region) and disomy 22.

In summary, regardless of alternate segregation, 6 disomy 22 products (seen in Table 1) could arise due to the segregation pattern of maternal translocation. However, our case is probably the only one that survived because trisomy on the short arm of Chr9 can be compatible with life.

Finally, as we mentioned in our article, the family was informed about all reproductive options, including alternate segregation (with normal and carrier fetuses) through an appropriate genetic counselling process, and preimplantation and prenatal diagnosis were recommended in subsequent pregnancies (1).

REFERENCES

1. Urtekin E, Kayhan G, Kazancioglu E, Yirmibes Karaoguz M. A partial trisomy 9 case with dicentric chromosome due to the adjacent-2 segregation of maternal reciprocal translocation. J Ist Faculty Med 2022;85(2):279-84. [CrossRef]
2. Meza-Espinoza JP, Picos-Cardenas VJ, Leal-Ugarte E. Comment on "A partial trisomy 9 case with dicentric chromosome due to the adjacent-2 segregation of maternal reciprocal translocation". J Ist Faculty Med 2024;87(3):265-6. [CrossRef]

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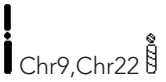
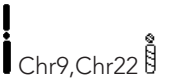
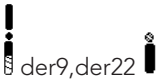
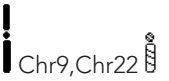
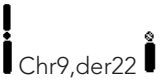
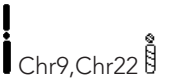
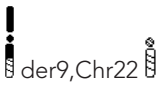
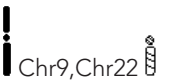
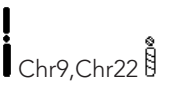
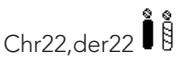
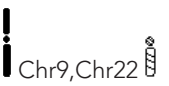
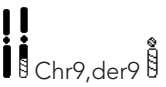
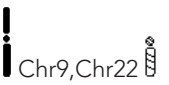
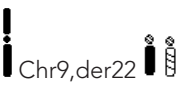
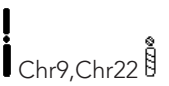
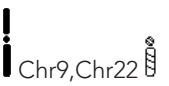
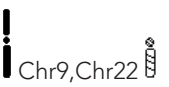
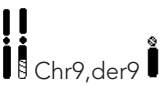
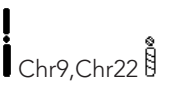
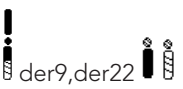
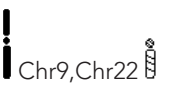
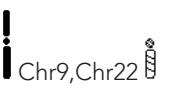
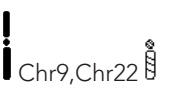
Table 1: The segregation patterns of translocation between chromosomes 9 (q22.31) and 22 (q11.1)

	MOTHER	FATHER	GAMETES
Alternate			Disomy 9–22
			Disomy 9–22 Balanced translocation carrier
Adjacent 1			Partial trisomy 9(q22.31 -> qter) and monosomy 22
			Partial monosomy 9(q22.31 -> qter) and trisomy 22
Adjacent 2			***Partial trisomy 9(pter->q22.31) and disomy 22****
			Partial monosomy 9(pter->q22.31) and disomy 22
Tertiary trisomy			Partial trisomy 9(pter->q22.31) and trisomy 22
			Partial trisomy 9(q22.31 -> qter) and **disomy 22****
Tertiary monosomy			Partial monosomy 9(pter->q22.31) and monosomy 22
			Partial monosomy 9(q22.31 -> qter) and **disomy 22****
Interchange Trisomy			Trisomy 9 and disomy 22
			Disomy 9 and trisomy 22
Interchange Monosomy			Monosomy 9 and disomy 22
			Disomy 9 and monosomy 22

*our case , **two cases of disomy 22 with partial trisomy/monosomy 9q