Polymorphisms of folate related genes and a risk of having a child with chromosome aneuploidy

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INTRODUCTION

Aneuploidy is the most common chromosome abnormality in humans and is the leading genetic cause of miscarriage and congenital birth defects. Several studies indicated that polymorphisms in folate-related genes can lead to DNA hypomethylation and abnormal chromosomal segregation.

AIM OF THE STUDY

The possible association of four polymorphisms in the three folate-related genes (MTHFR C677T, MTHFR A1298C, MTR A2756G and MTRR A66G) with a risk of having a fetus with chromosome aneuploidy was the aim of this study.

MATERIALS AND METHODS

We studied a total of 48 parents with a fetus with trisomy 21 (n=25), trisomy 18 (n=9), trisomy 13 (n=3), 45, X0 (n=5), 47, XXY (n=3) and triploidy (n=3). The parental origin of the aneuploidy was determined by QF-PCR analysis of several STR markers on each affected chromosome in the fetus and both parents. The MTHFR C677T, MTHFR A1298C, MTR A2756G and MTRR A66G were genotyped by multiplex PCR followed by SNaPshot analysis (Figure1).

RESULTS

The distribution of the allele and genotype frequencies of the four studied folate polymorphisms in the parents from whom the aneuploidy originated were compared with the frequencies in their partners as well as with ethnic matched individuals from the general population. Allele frequencies of four folate-related gene polymorphisms among patients and controls are presented in Figure 2, while the genotype frequencies are given in Table 1.

No statistically significant difference was observed in the allele and genotype frequencies of the four studied folate polymorphisms: MTHFR C677T, MTHFR A1298C , MTRR A66G and MTR A2756G between the parents from whom the aneuploidy originated and the control groups.

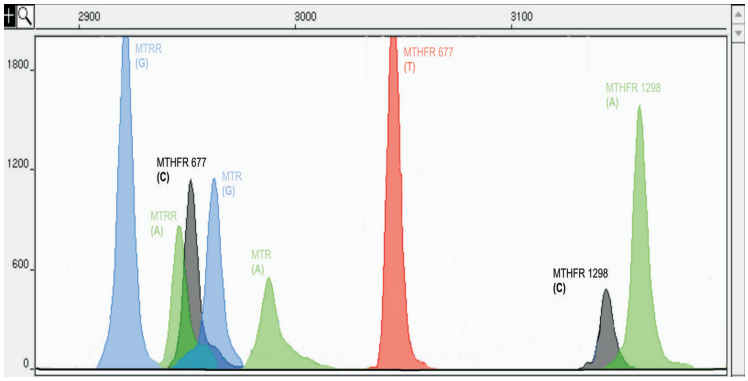
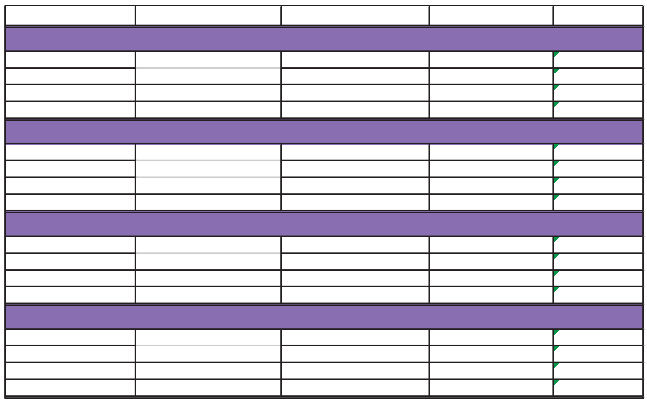


Figure 1. Electrophoreogram of the multiplex SNaPshot analysis for the detection of four folate-related genes polymorphisms .

Table 1. Genotype frequencies of four folate-related gene polymorphisms among parents from whom the aneuploidy originated and controls from the general population.



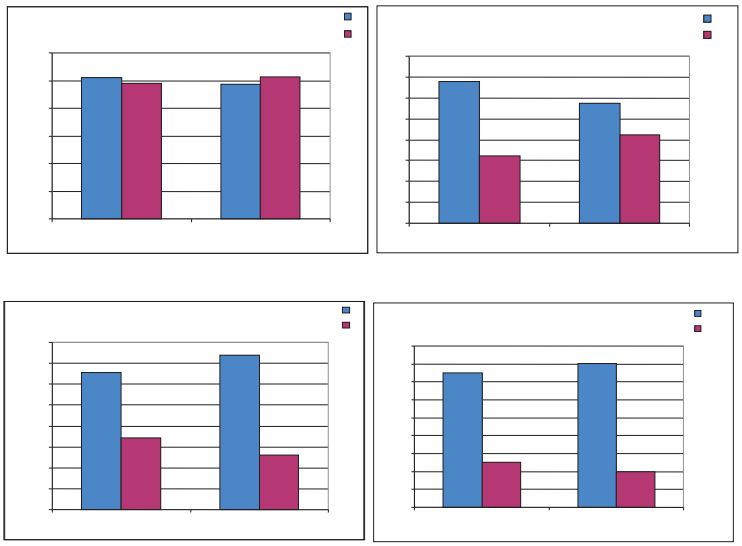


Figure 2. Allele frequencies of four folate-related gene polymorphisms among parents from whom the aneuploidy originated and controls from the general population.

CONCLUSIONS

Our study do not support the relationship between the four studied folate polymorphisms and a risk of having a child with an aneuploidy.

ACKNOWLEDGMENTS

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