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P134. The shift in emphasis of infertility genetic risk caused by defects in folate related gene polymorphism

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Context: Folate metabolism disorders and MTHFR gene polymorphism are the most studied issue in female and male infertility. The adverse effects of homocysteine and gene polymorphism on infertility risk have been proven in some studies and disproved in others. The ambiguous result these investigations may be due to an insufficient analysis of other folate genes polymorphism. The objective was to evaluate polymorphic variants frequency in folate related genes for patients from infertility and recurring early pregnancy loss couples compared to population study (1000 genomes). Methods: All patients underwent standard clinical examination, study of MTHFR (C677T, A1298C), MTRR (A66G), MTR1 (A2756G) genes polymorphism. Statistical analysis carried out using SPSS17.0. We compared genotypes and allele frequencies in all groups calculated HWE and ?2-criterion. To establish the risk of reproductive disorders develop was determined odds ratio (OR) and 95% confidence interval (CI) for dominant, recessive, co-dominant and additive genetic models. Participants: 685 patients were examined: 259 (195 women/64 men) with over 5 years infertility; 399 (290 women/109 men) with early reproductive losses. Interventions: We found no differences in the frequency of MTHFR variants (677CC: 43.92%, 40.56%; 677CT: 43.77%, 45.92%; 677TT: 12.31%, 13.52; 1298AA: 48.33%, 47.51%; 1298AC: 39.66%, 42.34%; 1298CC: 12.01%, 10.14%, respectively). Main outcome and measures: 66AA genotype of MTRR was significantly higher in population group (25.65% and 19.45% (?2= 5.99, p=0.014, OR=0.70 CI:0.53-0.92) The other MTRR genotypes frequencies did not differ (66AG: 47.87% , 44.14%; 66GG: 32.67%, 30.21%). 2756AA genotype of MTR1 gene was significantly increased in patients (57.29%, 45.81%, respectively (?2=14.38, p=0.001, OR=1.59 CI: 1.25-2.01). 2756AG and 2756GG genotypes frequencies was significantly higher in population group (44.17%, 10.02% compared to 36.47%, 6.23% (?2=6.62, p=0.010, OR=0.73 CI:0.57-0.92 and ?2=5.06, p=0.024, OR=0.60 CI:0.39-0.92, respectively). Significant genetic models association were dominant for MTRR gene (?2=6.34; p=0.01) and additive for MTR1 gene (?2=16.21; p=6.0E-5). Results: MTR1/ MTRR genotypes influenced on folate related one-carbon metabolism needed co-factor vitamin B12. Conclusion: Defined dominant MTRR and additive MTR1 genetic models associated with reproductive disorders shift the emphasis of further research on folate cofactors and DNA biosynthesis processes.

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