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SINGLE NUCLEOTIDE POLYMORPHISMS AS FACTORS IN THE FEMALE REPRODUCTIVE SYSTEM DISORDERS DEVELOPMENT

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Most disorders of the female reproductive system, such as infertility, habitual miscarriage, eclampsia, PCOS and endometriosis are classic examples of multifactor diseases. Hereditary component is expressed in the pathological alleles of single nucleotide polymorphisms (SNP) present in the genome. In present study, the analysis of our own data and literature about the role of the SNP of genes in the folic acid cycle, the factors of thrombophilia, the renin-angiotensin system, hormonal receptors and congenital immunity in the development of gynaecological disorders is made. More than 3000 cases of female reproduction violations associated with the presence of a possible hereditary component have been analyzed. It was demonstrated that one of the most important factors in the pregnancy violations development and failure in the use of assisted reproductive technologies, including IVF, is the presence of the folic acid cycle genes in various combinations in the genome of pathological alleles. Phenotypic manifestations of these features of the individuals' genotype are the general increase in the level of homocysteine in the blood (hyperhomocysteinemia), significant fluctuations of the homocysteine level values in time, disruption of global cellular regulatory mechanisms due to a change in the methylation pattern. The most important are SNP in MTHFR and MTRR genes. The polymorphisms in the genes MTR, SLC19A1 and MTHFD1 have much less significant effect. Others identified hereditary factors of the female reproductive system disorders development are the SNP pathological alleles in the thrombophilia factors genes. SNP in the genes FV (Leiden mutation), FII, PAI-I and ITGB3 showed the greatest value. Clinically significant phenotypic manifestation is expressed in the thrombus correct formation processes violation during the placenta vessels expansive developing. The role of polymorphisms in the genes of the innate immunity system carrying out intracellular signals and maintaining vascular tone is actively discussed. With reference to female reproduction system, the clinical significance of SNP in these gene networks seems not so great. As a result, the hypothesis about the special form of the cell regulatory systems responsible for the multifactorial diseases development is emerging. The peculiarities of the functioning of such systems called suertal require additional studies and discussions.

Biography

Andrei V Ivanov has completed his PhD with specialization in Cell Biology in 2007. He also has a specialization in Clinical Laboratory Diagnostics. Since 2009, he has been working in the Clinic of Advanced Medical Technologies named after N I Pirogov of Saint-Petersburg State University. Now, he is the Head of Department of Human Genetics. One of his most productive researches is the influence of genetic part in multifactorial diseases development such as female reproductive system disorders, irritable bowel syndrome, and metabolic syndrome. His area of interests includes metabolomics (especially steroid profiling), cell therapy of diabetic foot syndrome and biobanking. He considers his main task results of the biological research into broad medical practice.

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