venous thromboembolism and other associated gestational vascular complications like Recurrent Pregnancy Loss (RPL), preeclampsia, intrauterine growth restriction, and placental abruption during pregnancy. These complications are a major cause of maternal and fetal morbidity and mortality.

Aim of the study. This study focuses on the women who reported RPL, without any positive pregnancy and the identification of genetic factors that lead to the formation of thrombosis (F2 G20210A, F5 G1691A, MTHFR C677T, MTHFR A1289C, MTR A2756G, MTRR A66G), involved in fibrinolysis (PAI-1 4G/5G) and their association with primary female infertility.

Materials and methods. Research design was constructed as case-control type. The case group was represented by 44 patients with RPL, without any positive pregnancy, with normal karyotype, and lack of other causes (intrauterine infections, uterine pathology) responsible for the RPL. The control group included 57 patients with 2 positive pregnancies who did not receive anticoagulant treatment. The Odds Ratio (OR) was calculated for the case group and control group, at a 95% confidence interval, and p values <0,005 were considered statistically significant. OR>1 demonstrate a strong association between mutation and RPL, OR<1 show a weak association.

Results. We found that G1691A mutation in F5 gene encoding factor V (Leiden) (for heterozygous genotype OR=8,84; 95% CI; 1,02-76,42; p<0,05) and mutation G20210A in gene F2 encoding factor II (prothrombin), (for heterozygous genotype OR=7,18; 95% CI; 0,81-63,87; p<0,05), are major risk factors for RPL and primary female infertility. Carriers of the homozygous genotype after mutant allele were not determined in either group. The 4G/5G polymorphism of the PAI-1 gene, in this study was not associated with RPL and primary female infertility. Analysis of genes involved in folate cycle as MTHFR C677T mutation (OR=3,33; 95% CI; 1,37-8,09; p<0,05 for the heterozygous genotype and OR=3,73; 95% CI; 0,99-14,05; p<0,05 for the homozygous genotype after the mutant allele), MTR mutation A2756G (for the heterozygous genotype OR=2,91; 95%CI; 1,19-7,08; p<0,05 and for the homozygous genotype after the mutant allele OR=6,30; 95%CI; 1,17-34,03; p<0,05), MTRR mutation A66G (for the heterozygous genotype OR=2,40; 95%CI; 1,02-5,62; p<0,05 and for the homozygous genotype after the mutant allele OR=5,77; 95%CI; 0,99-33,68; p<0,05),demonstrated that these polymorphisms are major risk factors of RPL and primary female infertility. A1289C mutation of the MTHFR gene was not associated with RPL and primary female infertility.

Conclusion. According to the results of the study, it is recommended the genetic diagnosis of all patients with RPL, without organic or infectious causes, for detection of the genetic factors involved in hereditary thrombophilia.

Key words: hereditary thrombophilia, recurrent pregnancy loss, primary female infertility

316. CLINICAL AND CYTOGENETIC VARIATIONS IN MALE INFERTILITY CAUSED BY KLINEFELTER SYNDROME

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Introduction. Klinefelter's syndrome (KS) is the most common genetic cause of human male infertility characterized by gynecomastia, hypogonadism and azoospermia. About 80–90% of patients with Klinefelter's syndrome have an homogenous 47,XXY karyotype, the classic form of Klinefelter's syndrome. The prevalence of Klinefelter's syndrome is 1 in 700 men. Many patients with Klinefelter syndrome remain undiagnosed due to clinical variations.

Aim of the study. The purpose of this study is to establish the peculiarities of clinical and cytogenetic variations in male infertility caused by Klinefelter's syndrome
Materials and methods. A group of 75 men suspected with Klinefelter syndrome was clinically-genetically assessed during medical genetic counseling at the Center for Reproductive Health and Medical Genetics of the Institute of Mother and Child. Karyotyping of peripheral blood lymphocytes according to standard methods G was used for confirmation of diagnosis.
Results. The average age of patients with Klinefelter syndrome was 32.7, the main reason for consulting was infertility. The most common chromosomal abnormality diagnosed in the 35 patients with Klinefelter syndrome was homogeneous trisomy 47,XXY (30 cases - 85.7%), followed by mosaic form (47,XXY/46,XY: 3 case), polysomy X-Y (48,XXYY: 1 case and pentasomy - 49,XXXXY: 1 case). The main phenotypic aspects in men with KS were: hypogonadism, gynecomastia, azoospermia, decreased penis size, mental retardation, increase level of FSH. Most patients with Klinefelter syndrome were significantly taller than patients with normal karyotypes.
Conclusions. Medical genetic counseling and cytogenetic analyzes (karyotyping) are necessary for confirmation of clinical diagnosis in patients suspected with Klinefelter's syndrome.
Key words: Klinefelter syndrome, infertility, diagnosis, karyotype, cytogenetic testing

317. HUMAN CHIMERAS

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Introduction. The fundamentals of genetics states that every organism has one type of DNA that is present within any cell, and each cell of one body contains the same DNA. There is no law without any exception. Genetic chimeras are organisms that own two cell lines, meaning they have cells that contain different DNA. At first glance this phenomenon seems impossible, as nature accepts no mistake, but the existence of these organisms is undeniable. Chimeras live around us, and without an DNA test they cannot be spotted, as they present no specific or abnormal features. The frequency of chimerism is still unknown, as no one tests for it.
Aim of the study. The main goal of this research is to report an interesting and poorly known phenomenon, along with its particularity and recent scientific researches in the domain.
Materials and methods. For this research, scientific reports from various on-line platforms as CELL press, or PLOS Biology were used, along with other informative sources from internet.
Results. The result of this research is a short glance into the future of genetics, as the study of chimerism leads towards a new scientific branch that seems appealing to biologists, organ engineering.
Conclusions. Chimeras are one of the most marvelous examples of natural wonders, and understanding the way they are created, can lead humanity to the understanding of its own origin, the origin of life.
Key words: Chimera, Two cell lines, different DNA.