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PECULIARITIES OF PRENATAL DIAGNOSTICS IN THE FIRST TRIMESTER OF GESTATION IN WOMEN WITH COMPLICATED OBSTETRICAL ANAMNESIS

Abstract. *With the purpose to improve prenatal diagnostics in women with habitual miscarriage of pregnancy cytogenetic examination (karyotyping) has been carried out. The investigations conducted and results obtained stipulate the necessity to perform medical-genetic consulting with karyotyping in the 1st trimester of pregnancy enabling to improve early and effective prenatal diagnostics of congenital developmental fetal defects with an accurate prognosis concerning the possibility to prolong pregnancy and manage it.*
Key words: habitual miscarriage of pregnancy, karyotyping.

Introduction. In spite of introduction and fulfillment of state programs in the sphere of public health recent decades in Ukraine are characterized by a considerable decrease of health index among the population resulting in reduction of its number [2, 5, 7]. There are certain reasons to believe that such tendencies are associated with a negative course of genetic processes in the Ukrainian population [1, 3, 6]. Congenital pathology is one of the leading causes of disability and low quality of life of children and adults both in the developed and developing countries of the world [4, 8]. The world medical statistics estimates approximately 5% of newborns diagnosed with congenital pathology [9]. 30-50 infants among those 1000 live births are known to have certain congenital or hereditary diseases. It should be noted that approximately 30% of perinatal and neonatal mortality is caused by the pathology with dominating genetic component.

«Genetization» of medicine resulted in the development of molecular medicine initiating new tendencies in medical science, and one of them is predictive medicine. Contrary to therapeutic and even preventive medicine it should be considered as the first and the earliest stage of an active effect on the human organism with the purpose to correct potentially possible pathology in time [2]. Karyotyping is a cytogenetic method enabling to find deviations in the structure and number of chromosomes that can be a cause of infertility, congenital disease and birth of a sick child. There are two main especially important types of this examination in medical genetics: examination of chromosomes of patients' blood cells and prenatal karyotyping that is examination of the fetal chromosomes. Karyotype examination is carried out by means of cytogenetic and molecular-cytogenetic methods. The method enables to identify karyotype (peculiarity of structure and the number of chromosomes) by means of karyogram registration. Cytogenetic examination is carried out in a proband, his/her parents, relatives or fetus in case of suspected chromosome syndrome or other chromosome disorder.

Objective. With the purpose to improve prenatal diagnostics in women with complicated obstetrical anamnesis and habitual miscarriage of pregnancy in particular, a comprehensive examination has been conducted.

Materials and methods. At the initial stage of the investigation clinical-statistical analysis of case histories of women with habitual miscarriage of pregnancy was made (n=30, I group), the analysis of medical cards of practically healthy women was made for comparison (n=30, II group). Cytogenetic examination (karyotyping) of both groups was a final stage. Pregnant women of I and II groups differed considerably by age. The majority of women were from 21 to 30 years, although the

age of 33,3% of women with habitual miscarriage of pregnancy was more than 30. Considering the fact that occupation produces a certain effect on miscarriage of pregnancy we have examined peculiarities of working conditions and found that office workers constituted the most numerous group (46,7% and 43,3%), workers of the industrial and agricultural enterprises were on the second position (30,0% and 33,3%), housewives were the rest. Primary disorders of hypothalamic-pituitary regulation have been convincingly proved to play a certain role in the pathogenesis of habitual miscarriage of pregnancy, as a rule manifested by menstrual disorders. To specify the role of this factor in the development of the disease among patients of our region the character of menstrual function since the moment of menarche was studied. The age of menarche was found to range from 10 to 16 in both groups, an average index was 13,5±1,3. None of the women was found to have early menarche, although in 13,3% of women from I group sexual maturation delayed and the first menstruation occurred after 15 years of age.

Results. Analysis of the volume of menstrual blood loss found that in 73,3% of patients and 76,7% practically healthy women moderate menstruations occurred that was of no reliable difference. 23,3% women from II group had a tendency to excessive menstruation, while pregnant women with habitual miscarriage presented an opposite situation (26,7% of women indicated hypo- and oligomenorrhea, and irregular menstruations since the moment of menarche). Menstruations were painless in the majority of patients (86,7% and 83,3%, respectively), the rest of them were with painful syndrome, although these data did not differ reliably. Among previously experienced gynecological diseases women with habitual miscarriage of pregnancy suffered from exacerbation of salpingo-oophoritis, cervical erosion, although these findings also did not differ reliably in comparison with the data from II group. It is an interesting fact that polycystic ovary syndrome was found in 23,3% of women from I group without pregnancy, while none of the women from II group suffered from this nosology.

Discussion. Chronic extragenital diseases were found in 66,7% of women with habitual miscarriage of pregnancy and in 53,3% of pregnant women with uncomplicated anamnesis .it should be noted that the structure of extragenital pathology in both groups was different. Thus, practically healthy women in general suffered from chronic cholecystitis, gastroduodenitis, colitis, pyelonephritis, and pancreatitis. Women with habitual miscarriage of pregnancy since their childhood had been suffering from different degrees of obesity, chronic decompensated tonsillitis, vegetal-vascular disorders of different types (with prevailing hypertensive component), diffuse nontoxic goiter, varicose dilation of veins of the lower limbs.

Karyotyping was conducted for 30 pregnant women with complicated obstetrical anamnesis according to indications from their parents' side. Analysis of karyotyping results determined that all the examined people had a female karyotype – 46, XX. 30,8% of them had deviations in the structure of chromosomes. Changes in the structure of a short arm of the 9th chromosome (9p+) were found in 23,3% of women (46,XX,9qh+; 46,XX,9ph). Polyploid metaphase plates were found with the same frequency which is indicative of mitosis prophase pathology in the form of chromosome conjugation, which initiates multipolar mitosis at the stages of metaphase and anaphase. Polyploid multinuclear cells are formed in the result of pathology of telophase. In single cases the following pathologic karyotypes were found: 46,XX, 1qh; 46,XX, 13stk+s+; 46,XX, homolog heteromorphism; 46,XX, 21pstk, plate with translocation 2:13; 46,XX, 15stk+s+, 22pstk+s+.

Conclusion. Therefore, investigations conducted and results obtained stipulate the necessity to perform medical-genetic consulting with karyotyping in the 1st trimester of pregnancy enabling to improve early and effective prenatal diagnostics of congenital developmental fetal defects with an accurate prognosis concerning the possibility to prolong pregnancy and manage it.

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