

THE ROLE OF CYTOGENETIC PRENATAL DIAGNOSIS IN PREVENTION OF CHROMOSOMAL ABNORMALITIES

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Introduction: The diagnosis of foetal chromosomal abnormalities is carried out at the population level by cytogenetic prenatal diagnostic technologies (CPD). The CPD solve a significant amount of pressing problems and critical issues related to the prophylaxis of genetic pathologies and to the prevention of birth of children with numerical and structural chromosomal abnormalities. Therefore, CPD contribute to the detection of genetic pathologies and especially of chromosomal abnormalities in foetuses before their birth. The core and value of cytogenetic prenatal diagnosis is determined first of all by the information on foetal genotype and by prevention and avoidance of childbirth with genetic diseases. Among the CPD methods most commonly is indicated the amniocentesis, including the study of foetal karyotype from 16 to 18 w.g. The CPD methods are considered widely applied and safe tests, during which the geneticist, specialized in genetic counselling, informs the proband correctly, completely and understandably on the role, benefits, risk level, indications and contraindications of these investigations.

The scope of the work is to highlight the role of prenatal cytogenetic diagnosis for identification of chromosomal abnormalities in foetuses at early stages of intrauterine development and decrease the incidence of chromosomal abnormalities in newborns.

Materials and methods: The process of investigation included the prospective medical – genetic counselling with the purpose of identification of the target group consisting in 12 938 pregnant women of the risk-group, being prescribed to be examined in CRHMG from the Institute of Mother and Child, in the period 2005-2014. 4731 (36.6 ± 0.4%) of total amount of women had cytogenetic prenatal diagnosis (CPD): amniocentesis and chorionic villus biopsy. Patients were divided into two clinical batches: a) group I: 4731(36,6 ± 0,4%, p<0,001) - pregnant women from medium and high risk group; b) group II: 8207 (63,4 ± 0,4%, p<0,001) - pregnant women from low risk group.

Results: Those two groups were comparable in age, gestational period, degree of genetic risk. The age of women in high and medium genetic risk group was between 17 to 44 years old (average age 26,1 ± 5,3 years). The pregnancy term, when addressing to the geneticist, was between 6 to 22 w.g. (mean 14 ± 5.2 w.g).

In 4731 pregnant women who performed cytogenetic prenatal diagnosis, most frequent chromosomal abnormality were aneuploidies, including autosomal trisomies, the most common: Down syndrome accounted for 75 cases (0,18±1,6 %), Patau syndrome - 9 cases (0,06±0,2%) and Edwards syndrome - 20 cases (0,09±0,4%). Among the diagnosed gonosomale abnormalities, monosomy X was found in 8 cases (0,05±0,16%) and Klinefelter syndrome - 10 cases (0,07±0,21%). During 2008 - 2012 five fetuses (0,05±0,1%) were diagnosed prenatally with Triple X syndrome, 6 fetuses (0,05±0,13%) with triploidy and 31 fetuses (0,7±0,12%) with other structural chromosomal syndromes.

The research and evaluation of results of cytogenetic prenatal diagnosis – karyotyping, allowed prenatal diagnosis of chromosomal abnormalities in 164 cases, which was 0,3±3,5% of total amount of pregnant women who carried out DPC in concerned period.

In the situations when numerical or structural chromosomal abnormality is diagnosed with a prognosis poor for life or incompatible with life, a therapeutic abortion may be an option, which is legally justified but controversial ethically and morally. During the medical-genetic counseling these aspects were analyzed from all points of view, taking into account the vital prognosis and quality of life. A medical-genetic advice was provided for patients. The decision to keep or not the pregnancy, depended on the couple, on parents and/or the mother.

Conclusions:

1. The methods of cytogenetic prenatal diagnosis (fetal karyotyping) and medical-genetic counseling contribute to reducing the frequency of chromosomal abnormalities in newborns.

2. Due to CPD methods was possible to prevent the birth in 164 (0,3±3,5%) cases with chromosomal abnormalities, prenatally diagnosed until 21 weeks of gestation.

3. In the structure of fetal chromosomal abnormalities, diagnosed in fetus until 22 weeks of gestation, the most common is Down syndrome - 75 cases (0,18±1,6%), Edwards syndrome - 20 cases (0,09±0,4%) and structural chromosomal abnormalities - 31 cases (0,12±0,7%).

Diagnosis of chromosomal abnormalities, using all the spectrum of biotechnologies, should be performed in prenatal period [2, 6]. The essence and value of PCD is determined mainly by the information on the genotype and phenotypic manifestations in fetuses and preventing the birth of children with genetic diseases. In this context, significantly increased the role of medical-genetic counseling since it ensures informing and advising the patient and his family about the hereditary pathology that contribute to making certain decisions about genetic testing and reproductive health. Prenatal cytogenetic diagnosis procedures have allowed detection of chromosomal In prenatal diagnosis, prenatal chromosomal analysis is applied during the following conditions. Presence of structural chromosomal or genome abnormality in one of the parents. Previous child with de novo chromosomal aneuploidy or another genomic imbalance. Higher maternal ages. Understanding the role of chromosomal abnormalities in the pathogenesis of haematological malignancies led to the development of a selective treatment options and gives prognosis information [13, 16–20]. 1.2. FISH development. The development fluorescence in situ hybridization (FISH) technique increased the resolution of visualization of the chromosome rearrangements which is at the submicroscopic level [12, 21].