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LABORATORY METHODS FOR PRENATAL DIAGNOSIS

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This article is aimed at discussing prenatal diagnosis of fetal genetic abnormalities as an important step in the detection and prevention of birth defects and genetic syndromes. The authors show the multi-vector nature of this problem, which requires an integrated approach and the participation of a multidisciplinary team of specialists, such as gynecologists, radiologists, neonatologists, clinical geneticists, and pediatricians. Among the many methods that allow diagnosing congenital genetic pathology, this publication highlights fluorescent hybridization *in situ*. In comparison with other immunogenetic methods, this technique allows assessing the genetic status of an individual cell and detecting several etiopathogenetically significant abnormal cells among thousands of others with a normal genotype. This is its advantage over PCR, in which the DNA of all cells is mixed and the result is averaged. The article provides indicators for the selection of patients for screening for fetal pathology. Prenatal screening pathways, as nowadays in most countries consist of similar tests. This article is meant to be an introduction into more detailed ethical discussions about prenatal screening. A new approach of prenatal testing (PNT) will be useful given the currently available diagnostic tests. Genetic tests and general trend of individualization in healthcare policies are directions for establishing prenatal diagnosis with consideration of ethical policies.

Key words: pregnancy, prenatal diagnosis, fluorescent hybridization in situ

Prenatal diagnosis is important in detecting and preventing chromosomal and single gen disease. Steele and Breg in 1966 applied procedure for previously mentioned points. They show, that amniotic cells could be cultured and the chromosomes analyzed. About later chorionic villus sampling (CVS) technique for fetal genetic diagnosis was published in 1968. Advances since the mid-1980s have been the development of CVS procedures in the first trimester and also using recombinant DNA techniques for the diagnosis of single gene disorders [1, 21].

Prenatal diagnosis is divided in noninvasive and invasive testing.

Non-invasive testing consists of maternal blood assay, and ultrasonography.

Invasive testing includes chorionic villus sampling (CVS), amniocentesis and cordocentesis and infrequently embriyo- and fetal tissue biopsy.

Prenatal diagnosis had been performed by cytogenetics, molecular cytogenetics and molecular genetics techniques [3, 20].

Also, preimplantation embryo biopsy is useable technique for diagnosis of some genetic disorders [6, 11]. In this method, in vitro fertilization and embryo culture is followed by biopsy of one or two outer cells at the 6-10 cell stage from the embryonel development [4, 14]. In situ hybridisation is the technique used nowadays for diagnosis.

Prenatal diagnosis is useful in order to inform pregnant ladies about the risk for newborns defects or genetic disorders in their intrauterine development [9, 17]. In this way, prenatal diagnosis is important for the life of future newborn baby. There are various prenatal diagnosis methods for prevention of the genetic diseases.

In prenatal diagnosis, take part a multidisciplinary team consisting of specialists as obstetricians, radiologists, neonatologists, clinical geneticists, pediatres [2, 19].

Results of tests, are important also for candidate parents [7, 22].

From one medical perspective, it is known that prenatal diagnosis is divided in two categories of tests, concretly noninvasive and invasive [10, 24]. Non invasive tests – techniques consist of maternal blood sample, and imaging by ultrasonography or infrequently magnetic resonance imaging [12, 13]. Invasive tests – techniques consist of chorionic villus sampling (CVS), amniocentesis and cordocentesis, fetal tissue and embriyo biopsy.

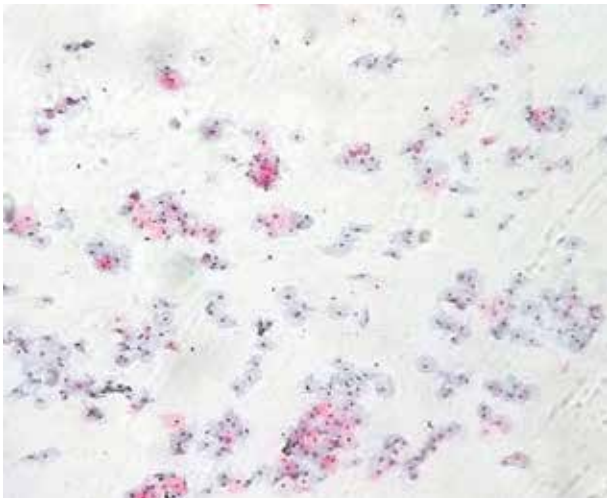

Figure 1 – Papanicolau smear.
Papanicolau stain x40 HPV

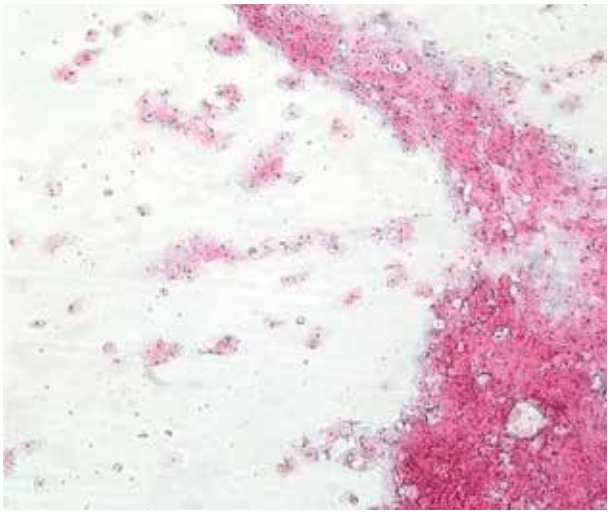

Figure 2 – Papanicolau smear.
Papanicolau stain. x40 Bacterian

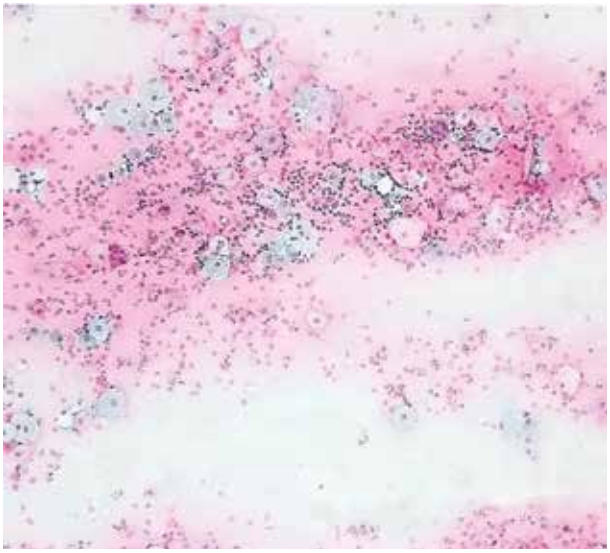

Figure 3 – Papanicolau smear.
Papanicolau stain x 40 Premalignancy

Screening method for detecting possible uterine cervix pathologies are also important to mention, in the context of the paper aim So, George Papanicolaou, together with Andromache Mavroyenous, his wife, discovered the screening test which is now recognized as the most significant advance in pathology and more than in the control of cancer nowadays [16, 23]. So the Papanicolaou smear, is known as a routine screening test for specific pathology especialy to detect malignancy as uterine cervix. This test was reported in 1928, and its efficacy was proved by 1941. For prenatal diagnosis is also useful this above mentioed test, in order to know the healthy status of the pregnant womens [18].

Indications for prenatal diagnosis

- Family history and maternal age.
- A positive screening test result is important.
- Structural fetal abnormality, structural parental chromosomal or genome abnormality detected by first trimester.
- Chromosomal aneuploidy or other genic and genomic imbalance in previous pregnancy [25].
- Methods for identify are imagistical and laboratory.

Fluorescence In Situ Hybridization technique **(FISH)**. FISH technique uses short sequences of DNA (probes) that carry fluorescent to detect complementary sequence of chromosomal DNA on cells and tisues [14]. Target region of FISH probe is metaphase chromosomes (metaphase FISH) or interphase nuclei (interphase FISH). Analyzis was performed by fluorescence microscopy with appropriate filters. CCD (charge-coupled device) camera based imaging and software programme are required for imaging of target region of applied FISH probe.

CONCLUSIONS

Prenatal screening pathways, as nowadays in most countries consist of similar tests. Our article is meant as an introduction into the more detailed ethical discussions about prenatal screening. A new approach of prenatal testing (PNT) will be useful knowing the curently diagnostic tests. For this purpose, could be including new techniques for risk-reducing, non-invasive sampling of foetal DNA and drastically enhanced possibilities of what may be faster analysed. Genetic tests and a general trend of individualization in healthcare policies are directions for establishing prenatal diagnosis with respect for ethical policies.

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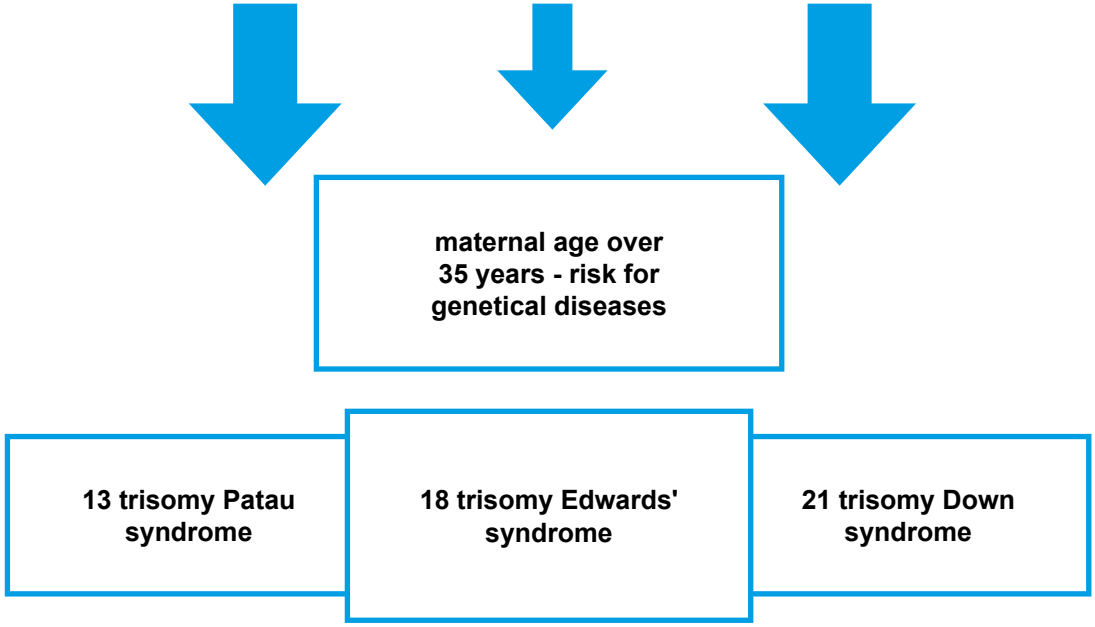

Figure 4 – Prenatal diagnosis points

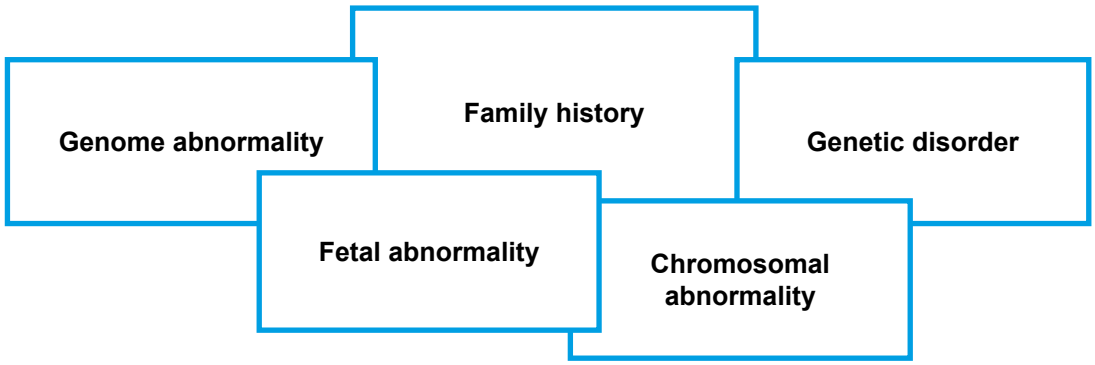

Figure 5 – Prenatal diagnosis points

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ЛАБОРАТОРНЫЕ МЕТОДЫ ПРЕНАТАЛЬНОЙ ДИАГНОСТИКИ

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Статья посвящена пренатальной диагностике генетических аномалий плода, как важному этапу для выявления и предотвращения врожденных дефектов и генетических синдромов. Авторами показана многовекторность данной проблемы, что требует комплексного подхода и участия мультидисциплинарной команды специалистов, как гинекологов, рентгенологов, неонатологов, клинических генетиков, педиатров. Среди множества методов позволяющих диагностировать врожденную генетическую патологию в данной публикации выделена флуоресцентную гибридизация (ФГ) in situ. В сравнении с другими иммуногенетическими методами данная методика позволяет оценить генетический статус отдельной клетки и обнаружить несколько этиопатогенетически значимых аномальных клеток среди тысяч других с нормальным генотипом. В этом его преимущество в сравнении с ПЦР, при котором ДНК всех клеток смешивается и результат усредняется. В статье даны индикаторы при отборе пациентов для проведения скрининга на патологию плода. В настоящее время траектория пренатального скрининга в большинстве стран, состоит из аналогичных тестов. Идея нашей статьи как введение в более подробные этические дискуссии о пренатальном скрининге. Новый подход к пренатальному тестированию (ПНТ)

будет полезен, зная современные диагностические тесты. Генетические тесты и общая тенденция к индивидуализации в политике здравоохранения являются направлениями для установления пренатальной диагностики с соблюдением этических норм

Ключевые слова: беременность, пренатальная диагностика, флуоресцентная гибридизация

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ТУА БІТКЕН ПАТОЛОГИЯЛЫҚ АУРУЛАРДЫ ДИАГНОСТИКАЛАУДЫҢ ЗЕРТХАНАЛЫҚ ӘДІСТЕРІ

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Бұл ғылыми мақала ұрықтың генетикалық патологиясын зертханалық диагностикалау туралы, туа біткен ақаулар мен генетикалық синдромдарды анықтау және алдын алудағы маңызды қадам ретінде. Авторлар кешенді тәсілді және гинекологтар, рентгенологтар, неонатологтар, клиникалық генетиктер және педиатрлар сияқты мамандардың көп салалы тобының қатысуын талап ететін бұл мәселенің жанжақтылығын көрсетті. Туа біткен генетикалық патологияны диагностикалауға мүмкіндік беретін көптеген әдістердің ішінде бұл басылым флуоресцентті ubгибридизация (ФГ) in situ көрсетеді Басқа иммуногенетикалық әдістермен салыстырғанда бұл әдіс жеке жасушаның генетикалық жағдайын бағалауға және қалыпты генотипі бар мыңдаған басқалардың арасында этиопатогенетикалық маңызды бірнеше анормальды жасушаларды анықтауға мүмкіндік береді. Бұл оның ТПР-дан артықшылығы, онда барлық жасушалардың ДНҚ-сы араласып, нәтиже орташа алынады. Мақалада ұрықтың патологиясын анықтау үшін скринингке пациенттерді таңдау көрсеткіштері берілген. Қазіргі уақытта көптеген елдерде пренатальды скрининг траекториясы ұқсас сынақтардан тұрады. Біздің мақаламыздың идеясы пренатальды скрининг туралы толығырақ этикалық талқылауларға кіріспе болып табылады. Қазіргі заманғы диагностикалық сынақтарды білу пренатальды тестілеуге (ПНТ) жаңа көзқарасты анықтайды. Генетикалық тесттер және денсаулық сақтау саясатын жекелендірудің жалпы тенденциясы этикалық түрде пренаталдық диагнозды орнату үшін қажет.

Кілт сөздер: жүктілік, пренатальды диагностика, флуоресцентті гибридизация