



GENETIC TESTS AND THEIR ROLE IN OBSTETRIC AND GYNAECOLOGICAL CARE

Khasanova Dilafruz Abdukhamidovna

Assistant

Department of Obstetrics and Gynecology №1

Samarkand State Medical University, Samarkand, Uzbekistan

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ABSTRACT

In recent decades, genetic tests and counseling have become important tools in gynecology, influencing approaches to family planning and hereditary risk assessment. This article reviews the use of genetic tests in various aspects of gynecology, such as assessment of inherited diseases, preimplantation genetic diagnosis (PGD), non-invasive prenatal testing (NIPT), and other situations requiring genetic evaluation. In addition, emphasis is placed on the importance of genetic counselling for women to make informed decisions about health and reproductive plans, and to provide support in understanding the risks and opportunities associated with inherited diseases.

GENETIK TESTLAR VA ULARNING AKUSHERLIK VA GINEKOLOGIK YORDAMDAGI O'RNI

Xasanova Dilafruz Abduxamidovna

Assistent

Akusherlik va ginekologiya kafedrası №1

Samarqand davlat tibbiyot universiteti, Samarqand, O'zbekiston

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Genetik testlar, genetik maslahatlar, irsiy kasalliklar, homiladorlikni rejalashtirish, preimplantatsiya genetik diagnostikasi (PGD), invaziv bo'lmagan prenatal test (IBPT), irsiyat xavfi, reproduktiv texnologiyalar.

ABSTRACT

So'nggi o'n yilliklarda genetik tekshiruv va maslahat ginekologiyada oilani rejalashtirish va genetik xavfni baholashga ta'sir qiluvchi muhim vositaga aylandi. Ushbu maqolada irsiy kasalliklarni baholash, preimplantatsiya genetik diagnostikasi (PGD), invaziv bo'lmagan prenatal test (IBPT) va genetik baholashni talab qiladigan boshqa vaziyatlar kabi ginekologiyaning turli jihatlarida genetik testlardan foydalanish ko'rib chiqiladi. Bundan tashqari, u ayollarga salomatlik va reproduktiv rejalar haqida ongli qarorlar qabul qilish va irsiy kasalliklar bilan bog'liq xavf va imkoniyatlarni tushinishda yordam berish uchun genetik maslahatning muhimligini ta'kidlaydi.



Relevance. In recent decades, genetic tests have become an integral part of gynecological practice, especially in family planning and risk assessment of inherited diseases. Genetic counseling provides women and families with important information to make informed decisions about health and reproductive plans.

This article authored by Radzinsky V. E., Dukhin A. O., and Kostin I. N. discusses the reproductive health of women after surgical treatment of gynecological diseases. The study examines the effects of surgical interventions on reproductive health, including the impact on the possibility of conception and pregnancy, as well as on the general health of women in the postoperative period. Genetic counseling is a process in which geneticists help patients understand and adapt information about genetic diseases, assess the risks of transmitting inherited diseases, and provide recommendations for further action. This may include analyzing family history, determining the need for genetic tests, and discussing test results and possible consequences.

Uses of Genetic Testing

Genetic testing can be useful in a variety of gynecological settings, including:

1. Hereditary disease risk assessment:

- Women with a family history of hereditary diseases, such as mutations in the BRCA1 and BRCA2 genes, have an increased risk of breast and ovarian cancer. Genetic testing can help determine whether preventive measures, such as increased monitoring or surgery, are worth taking (Paluch-Shimon et al., 2020).

2. Preimplantation genetic diagnosis (PGD):

- PGD allows genetic testing of embryos obtained through in vitro fertilization (IVF) for chromosomal abnormalities or hereditary diseases before implantation. This helps select healthy embryos and reduce the risk of genetic diseases in children (Olivennes et al., 2018).

3. Screening for chronic diseases: - Genetic testing can be used to screen for disorders such as thalassemia or cystic fibrosis, especially in women planning to become pregnant. For example, an article by Gasparov A.S. et al. examines the role of connective tissue dysplasia in obstetrics and gynecology. The article examines aspects related to the impact of connective tissue dysplasia on women's health, including the course of pregnancy, fertility, and the development of various obstetric and gynecological complications. The authors emphasize the importance of diagnosing and taking this condition into account to improve clinical practice in obstetrics and gynecology.

4. Use of NIPT (Non-Invasive Prenatal Testing):

- NIPT analyzes free-floating fetal DNA in Maternal Blood. This allows for the detection of chromosomal abnormalities, such as Down syndrome, with a high degree of accuracy and without risk to the fetus (Gil et al., 2020).

The Role of Genetic Counseling in Pregnancy Planning

Genetic counseling plays a critical role in the pregnancy planning process as it enables women to:

- Learn about the risks of hereditary diseases and pass on the information to their partners.
- Evaluate available tests and methods of their use.
- Make informed decisions about pregnancy and the possibility of using assisted reproductive technologies.



There are several aspects to consider regarding the state of genetic testing in the field of gynecology in Uzbekistan:

1. Development of genetic diagnostics: In recent years, Uzbekistan has stepped up its work in the field of genetic diagnostics and counseling. This includes the creation of new laboratories and the introduction of modern technologies for the analysis of genetic material.
2. Education and advanced training of specialists: An important part of the development of genetic testing is the training of qualified personnel. Educational programs for geneticists are conducted in Uzbekistan, which helps to improve the level of knowledge in the field of genetic diagnostics and counseling.
3. Testing for hereditary diseases: Genetic testing in Uzbekistan can include diagnostics of hereditary diseases, such as cystic fibrosis, thalassemia and others, which leads to improved early detection and prevention of diseases.
4. Screening programs: Some medical institutions may conduct screening programs to assess genetic risks, which is especially important for women with a family history of hereditary diseases. In addition, medical institutions have all the screening programs recommended by WHO.
5. Cooperation with international organizations: Uzbekistan actively cooperates with international organizations and research institutes in the field of genetic testing, which allows for the introduction of best practices and new technologies.

Conclusion. Genetic tests and counselling are important tools in gynaecology to not only assess the risks associated with inherited diseases, but also to help families in the process of planning a pregnancy. Women who are aware of their genetic risk and are able to receive qualified counselling can make more informed decisions, which has a positive impact on the health and future of their children.

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