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PREIMPLANTATION GENETIC TESTING FOR ANEUPLOIDY (PGT-A) AND NON-INVASIVE PRENATAL TESTING (NIPT) AS KEY COMPONENTS OF CONTEMPORARY PREGNANCY MANAGEMENT

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Abstract:
The rapid development of reproductive genetics has substantially influenced current approaches to pregnancy planning and prenatal care. Preimplantation genetic testing for aneuploidy (PGT-A) and non-invasive

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prenatal testing (NIPT) are innovative screening methods designed to identify chromosomal abnormalities at different stages of human development. PGT-A is performed prior to embryo implantation during assisted reproductive technology cycles, while NIPT allows early assessment of fetal chromosomal status through maternal blood analysis. The combined use of these methods contributes to improved reproductive outcomes, reduction of pregnancy loss, and optimization of individualized pregnancy management. This article provides an analytical overview of the principles, clinical significance, advantages, limitations, and ethical aspects of PGT-A and NIPT in modern obstetric practice.

Keywords: In vitro fertilization, pregnancy management, diagnostic challenges, reproductive outcomes.

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Materials and Methods:

Chromosomal abnormalities remain a major cause of failed implantation, spontaneous miscarriage, and congenital disorders. The prevalence of aneuploid embryos increases significantly with advancing maternal age, which has become a global trend due to delayed childbearing [1]. As a result, modern obstetrics increasingly relies on genetic screening technologies to improve pregnancy outcomes and reduce perinatal morbidity.

Congenital anomalies affect millions of newborns worldwide each year and represent a serious public health challenge [2]. Conventional invasive prenatal diagnostic procedures, although highly informative, are associated with procedural risks and psychological stress for pregnant women. Therefore, the introduction of non-invasive and preimplantation screening strategies has become a priority in contemporary pregnancy management.

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PGT-A and NIPT represent two complementary approaches that allow assessment of chromosomal integrity before and during pregnancy. Their integration into clinical protocols supports personalized medicine, informed decision-making, and evidence-based reproductive care [3]. This study is based on an extensive review of international scientific publications from 2010 to 2024. Data were collected from major medical databases and peer-reviewed journals, including Fertility and Sterility, Human Reproduction, The New England Journal of Medicine, and American Journal of Obstetrics and Gynecology.

The analysis included randomized controlled trials, systematic reviews, meta-analyses, and clinical guidelines focusing on diagnostic accuracy, pregnancy outcomes, and clinical implementation of PGT-A and NIPT. Ethical considerations and limitations reported in the literature were also evaluated.

Preimplantation Genetic Testing for Aneuploidy: Clinical Principles

PGT-A is performed during in vitro fertilization cycles and involves genetic analysis of embryos prior to uterine transfer. Embryo biopsy is typically carried out at the blastocyst stage, followed by chromosomal assessment using advanced genomic technologies such as next-generation sequencing [4].

The primary clinical objective of PGT-A is to identify embryos with a normal chromosomal complement, thereby increasing the likelihood of successful implantation and reducing the risk of miscarriage. This method is particularly beneficial for women of advanced reproductive age, couples with recurrent pregnancy loss, repeated implantation failure, and certain male factor infertility cases [5].

Although numerous studies report improved implantation rates and reduced pregnancy loss following PGT-A, challenges such as embryonic

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mosaicism and interpretation of genetic results remain areas of ongoing research [6].

Non-Invasive Prenatal Testing: Methodology and Applications

NIPT is based on the detection of cell-free fetal DNA fragments circulating in maternal plasma.

This screening test can be performed early in pregnancy and demonstrates high sensitivity for common chromosomal abnormalities, including trisomies 21, 18, and 13 [7].

The non-invasive nature of NIPT eliminates the risk of procedure-related pregnancy loss and has led to its widespread adoption in prenatal screening programs, particularly among high-risk populations [8]. In addition to autosomal aneuploidies, NIPT can provide information on sex chromosome abnormalities and fetal sex.

Despite its high diagnostic performance, NIPT remains a screening tool rather than a definitive diagnostic test. Factors such as low fetal DNA fraction and placental mosaicism may affect test accuracy, necessitating confirmatory invasive testing in cases of positive results [9].

Integration of PGT-A and NIPT in Pregnancy Management

The combined application of PGT-A and NIPT enables a continuous genetic assessment pathway from embryo selection to early prenatal screening. In assisted reproductive technology cycles, embryos selected through PGT-A may subsequently be monitored using NIPT, providing an additional layer of genetic risk evaluation [10].

This integrated approach contributes to improved clinical outcomes, including lower miscarriage rates, reduced incidence of chromosomal abnormalities at birth, and enhanced patient reassurance. Furthermore, it supports individualized counseling and optimized obstetric management strategies.

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Advantages and Limitations

Advantages

- Improved embryo selection and implantation success
- Early identification of chromosomal abnormalities
- Reduction in invasive diagnostic procedures
- Enhanced pregnancy outcomes and patient satisfactio

Limitations

- High cost and limited availability in some healthcare systems
- Ethical concerns related to embryo selection
- Diagnostic challenges associated with mosaicism
- Need for confirmatory invasive testing following abnormal NIPT results

These factors highlight the importance of appropriate patient selection and comprehensive genetic counseling [11].

Ethical and Clinical Considerations

The increasing use of genetic screening technologies raises important ethical issues, including reproductive autonomy, informed consent, and the potential psychological impact on patients. Healthcare providers must ensure that patients receive balanced and accurate information regarding the benefits and limitations of PGT-A and NIPT

Ethical practice requires that genetic testing be offered within a framework that respects patient values, cultural considerations, and evidence-based clinical guidelines [12].

Conclusion:

PGT-A and NIPT have become integral components of modern pregnancy management, offering powerful tools for reducing the burden of

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chromosomal abnormalities and improving reproductive outcomes. Their combined use supports early risk assessment, personalized clinical care, and informed decision-making.

While technological and ethical challenges remain, continued research and refinement of clinical protocols will further enhance the role of these methods in obstetrics and gynecology. The appropriate integration of PGT-A and NIPT represents a significant step toward safer and more effective pregnancy management.

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