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DNK DIAGNOSTICS AND GENETIC SCREENING IN MODERN MEDICINE

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Annotation: This article will highlight the scientific and practical importance of DNK Diagnostics and genetic screening in modern medicine. Thanks to advances in Molecular Biology, the possibilities of detecting genetic diseases, diagnosing them at an early stage and individual treatment are expanding. The study examined DNK analysis methods — polymerase chain reaction, high conductive sequencing, microarray technologies, and the effectiveness of prenatal and neonatal screening programs. The clinical applications of these processes, their role in the health care system, economic and ethical aspects were also analyzed. The results obtained show that the widespread introduction of genetic screening programs is important in improving the general health of the population, treating diseases at an early stage and developing the concept of individual medicine.

Keywords: DNK diagnostics, genetic screening, molecular diagnostics, medical genetics, prenatal screening.

Introduction

Genetic disorders and DNK-level changes are essential factors in modern medicine for diagnosing diseases, planning prevention strategies, and developing individualized therapies. Alongside advancements in molecular biology and genomics, DNK diagnostics have enabled precise and rapid detection. Genetic screening programs are used to identify disease risks at early stages within populations, guide reproductive decisions, and ensure the birth of healthy generations. The purpose of this article is to analyze DNK diagnostic and genetic screening methods, assess their effectiveness, and highlight their role in clinical practice.

Methods

This research is based on methods for studying scientific literature, analyzing existing national and international recommendations, and comparing technologies in the field of molecular diagnostics. Key techniques include polymerase chain reaction (pzr), real-time PZR (qPCR), Sanger sequencing, highly conductive sequencing (NGS), microarray analysis, and genotyping.his research is based on methods for studying scientific literature, analyzing existing national and international recommendations, and comparing technologies in the field of molecular diagnostics. Key techniques include polymerase chain reaction (pzr), real-time PZR (qPCR), Sanger sequencing, highly conductive sequencing (NGS), microarray analysis, and genotyping. Prenatal screening (NIPT — non-positive prenatal testing) as well as newborn

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screening experiments were also examined. Clinical examples and results from practice were also summarized throughout the work.

Results

Capabilities of molecular diagnostic methods:

PCR and qPCR allow for the rapid and highly sensitive detection of specific mutations.

Sanger sequencing is considered the gold standard for confirming small variations.

NGS enables the analysis of the entire genome or gene panels within a short time, identifying a wide range of pathogenic variants.

Effectiveness of screening programs:

NIPT provides highly accurate diagnosis of chromosomal abnormalities and reduces the need for invasive tests (amniocentesis, chorionic villus sampling).

Newborn screening enables the early detection of metabolic and hormonal disorders, allowing timely application of treatment and preventive measures.

Examples of clinical applications:

In oncology, identifying tumor-specific genetic parameters has improved the ability to select targeted therapies (e.g., HER2, EGFR, BRAF mutations, and others).

In pharmacogenetics, individualizing drug dosages and selections helps reduce adverse drug reactions.

Debate

DNK Diagnostics and genetic screening have many advantages in clinical practice: accuracy, early diagnosis, individual therapy, and preventive measure planning. However, there are also restrictions related to this area — technical restrictions, options that are lost or incorrectly interpreted, as well as financial issues. Despite the fact that NIPT is highly sensitive, it is not diagnostic — confirming invasive tests may be needed.ebate

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Ethical aspects: genetic information is personal and sensitive. Issues of preservation, privacy, and protection against genetic discrimination are important. The genetic counseling service helps patients explain outcomes, assess risks, and make decisions.

Impact on the health system: the widespread introduction of genetic screening requires funding, training and technological infrastructure. But in the long run, these programs can reduce health care costs by detecting diseases early.

Conclusion

DNK Diagnostics and genetic screening have taken a central place in modern medicine, expanding the possibilities of personal medicine and early prevention. Molecular methods are effective in identifying diseases and individualizing therapy, and screening programs promote improved public health. However, given the technical, economic and ethical limitations, it is necessary to responsibly introduce these technologies.onclusion

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