E CONF SERIES

International Conference on Economics, Finance, Banking and Management
Hosted online from Paris, France
Website: econfseries.com 24th June, 2025

LABORATORY DIAGNOSIS OF HEREDITARY DISEASES: A REVIEW OF MODERN METHODS AND THEIR CLINICAL SIGNIFICANCE

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Annotation
Hereditary diseases are a group of pathologies caused by mutations in the genetic material transmitted by inheritance. Timely and accurate laboratory diagnosis of such diseases is crucial for early intervention, prognosis and genetic counselling. This review article focuses on modern methods of laboratory diagnosis of hereditary diseases, including molecular genetic, biochemical and cytogenetic approaches, and their integration into clinical practice [5,6,7].

Keywords: hereditary diseases, chronic diseases, molecular biology, biochemical methods, diagnosis;

Introduction

Hereditary disorders constitute a significant proportion among congenital and chronic pathologies, especially in paediatric and genetic practice. According to WHO, about 5% of newborns suffer from various forms of inherited disorders. Due to progress in molecular biology and bioinformatics, the possibilities of early diagnosis have significantly expanded, which allows not only to improve the quality

of life of patients, but also to implement prenatal and neonatal screening strategies [8,9,10].

Classification of laboratory diagnostic methods. Methods of laboratory diagnostics of hereditary diseases can be conditionally divided into several main groups [21,22,23,24]

Polymerase chain reaction (PCR) can detect point mutations, deletions, and insertions in specific regions of DNA.

- **o DNA sequencing, including Sanger and high-throughput sequencing (NGS),** is used to analyse single genes as well as whole exomes or genomes**.**

2. **Fluorescence** in situ hybridisation (FISH) is used to detect chromosomal rearrangements and microdeletions.

3. **Microarray technology** provides simultaneous testing of multiple genetic markers.

1. Cytogenetic Methods

2. **Karyotyping allows** the determination of numerical and structural abnormalities of chromosomes.

4. **High-resolution** cytogenetic analysis is used when subtelomeric rearrangements are suspected**.**

5. Biochemical methods

Used in inherited metabolic disorders such as phenylketonuria, galactosemia, and accumulation diseases.

Metabolite levels (e.g. amino acids, organic acids) are analysed in serum, urine and liquor.

Platforms such as tandem mass spectrometry (MS/MS) are used.

The role of neonatal screening. Neonatal screening is a public programme for the early detection of a number of inherited diseases. In most countries, it includes the detection of pathologies such as hypothyroidism, phenylketonuria, cystic fibrosis and adrenogenital syndrome. The use of highly sensitive laboratory methods makes it possible to diagnose the disease before symptoms appear and to start therapy in a timely manner [11,12,13,14,15]**.**

Genetic counselling and ethical considerations. The results of laboratory diagnosis of inherited diseases play an important role in the genetic counselling

process. Detection of pathogenic mutations in a patient or carriers in parents requires a sensitive approach and ethical standards, including confidentiality, informed consent and the right to not know the diagnosis.The role of neonatal screening. Neonatal screening is a public programme for the early detection of a range of inherited diseases. In most countries, it includes the detection of pathologies such as hypothyroidism, phenylketonuria, cystic fibrosis and adrenogenital syndrome. The use of highly sensitive laboratory methods makes it possible to diagnose the disease before symptoms appear and to start therapy in a timely manner [16,17,18,19,20].

Conclusion

Laboratory diagnosis of inherited diseases is an integral part of modern medicine, providing the basis for personalised therapy, reproductive choice and prevention. Advances in technologies such as NGS and bioinformatics analysis continue to transform diagnostic approaches, making them more accurate, accessible and informative. However, along with technical progress, issues of standardisation, data interpretation and interdisciplinary collaboration between specialists remain relevant.

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