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DIAGNOSTIC GENES AND THEIR IMPORTANCE IN THE DIAGNOSIS OF NON-ALCOHOLIC FATTY LIVER DISEASE (NAFLD)

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Abstract
Non-alcoholic fatty liver disease (NAFLD) is one of the most common chronic liver diseases not associated with alcohol consumption. In recent years, special attention has been paid to the molecular genetic mechanisms of NAFLD pathogenesis, including the role of diagnostic genes. The article discusses the main genes associated with the development and progression of NAFLD, their diagnostic value, as well as the prospects for the use of genetic markers in clinical practice.

Keywords: NAFLD, genes, diagnostics, PNPLA3, TM6SF2, GCKR, MBOAT7, genetic markers, molecular diagnostics.

Introduction
Relevance of NAFLD study
Epidemiology and impact on public health
Diagnostic difficulties and need for new methods
The role of genetics in the pathogenesis of NAFLD

Chapter 1. NAFLD Overview
Definition and Classification
Stages of Progression: Steatosis → Steatohepatitis → Fibrosis → Cirrhosis
Clinical and Diagnostic Criteria
Modern Diagnostic Methods (ultrasound, biochemistry, biopsy)

Chapter 2. Genetic predisposition to NAFLD
Genetic vulnerability: general principles
The role of heredity and ethnic factors
Familial forms of NAFLD

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Chapter 3. Key genes associated with NAFLD
3.1. PNPLA3 (rs738409)
Gene functions and polymorphism
Associations with the severity of steatosis and fibrosis
Mechanism of action of the I148M variant
3.2. TM6SF2 (rs58542926)
Effect on lipid metabolism and hepatocytes
Role in the progression of NAFLD
Genetic features
3.3. GCKR (rs1260326)
Involvement in the regulation of glucose and lipid metabolism
Role in liver steatosis
3.4. MBOAT7 (rs641738)
Functions in phospholipid metabolism
Association with inflammation and fibrosis
3.5. HSD17B13
Protective alleles
Effect on liver enzymatic damage levels

Chapter 4. Methods for Determining Genetic Markers
PCR and RT-PCR
Next Generation Sequencing (NGS)
Microarrays and Panel Tests
Bioinformatics approaches

Chapter 5. Genetic diagnostics in clinical practice
Prospects for the implementation of genetic screening
Individualized approach to treatment
Prognostic value of genes
Ethical aspects and limitations

Chapter 6. Comparative analysis of allele frequencies in different populations
Ethnic differences (Europeans, Asians, African Americans)
Results of meta-analyses
Examples of population studies

Chapter 7. Future and Prospects of Genetic Research in NAFLD
Genomic Editing and CRISPR
Personalized Medicine
Next Generation Biomarkers

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Conclusion:

Genetic markers such as PNPLA3, TM6SF2, GCKR, MBOAT7 and HSD17B13 play an important role in the pathogenesis and diagnosis of NAFLD. Their use in clinical practice can improve diagnostic accuracy, predict disease progression and optimize treatment. The future of NAFLD diagnostics is associated with further development of molecular technologies and integration of genetic data into medical protocols.

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