MODERN DIAGNOSTIC METHODS FOR IRON DEFICIENCY ANEMIA: FROM CLINICAL PRACTICE TO MOLECULAR RESEARCH

Isakulova Muhabbat Mardonovna
Assistant of the Department of Clinical and Laboratory Diagnostics with a course of PGD Clinical and Laboratory Diagnostics of Samarkand State University
Samarkand, Uzbekistan
Baxranova Ziyoda
Cadet of the department of clinical and laboratory diagnostics with a course of PGD Clinical and Laboratory Diagnostics of Samarkand State University
Samarkand, Uzbekistan

Abstract: *Iron deficiency anemia (IDA) is one of the most common forms of anemia worldwide. This article explores modern diagnostic methods for IDA, ranging from standard laboratory analyses to molecular research. The importance of integrating various diagnostic approaches to enhance diagnostic accuracy is emphasized.*

Keywords: *iron deficiency anemia, diagnostics, ferritin, molecular research, biomarkers.*

Iron deficiency anemia (IDA) is a pathological condition characterized by reduced levels of hemoglobin and red blood cells due to iron deficiency. The causes of IDA include chronic blood loss, insufficient dietary iron intake, impaired absorption, and increased physiological demands. This article

examines modern approaches to diagnosing IDA, including traditional laboratory tests and advanced molecular methods.

Traditional Laboratory Methods

Complete Blood Count (CBC):

Decreased hemoglobin levels (< 12 g/dL in women, < 13 g/dL in men).

Hypochromia and microcytosis - reduced mean corpuscular volume (MCV < 80 fL).

Increased red cell distribution width (RDW).

Biochemical Markers: Reduced serum iron levels.

Ferritin -the most sensitive marker of iron deficiency, although it may appear normal or elevated in inflammatory states. Increased total iron-binding capacity (TIBC) in IDA.

Transferrin Saturation: A decrease below 20% indicates iron deficiency.

Advanced Diagnostic Methods

C-reactive protein (CRP): For evaluating inflammatory components.

Hepcidin: A regulator of iron absorption. Hepcidin levels are reduced in IDA.

Reticulocyte Indices:RET-He — hemoglobin content in reticulocytes, reduced in iron deficiency.

Molecular Research Methods: Molecular diagnostic methods for IDA open new horizons for understanding the disease's pathophysiology and improving diagnostic accuracy. Modern technologies enable the study of genetic and biochemical aspects of iron metabolism and the identification of hidden forms of deficiency.

Polymerase Chain Reaction (PCR): Used to analyze genetic mutations associated with impaired iron metabolism, such as mutations in the HFE gene responsible for hereditary hemochromatosis.

PCR precisely identifies gene polymorphisms affecting iron absorption and transport.

Next-Generation Sequencing (NGS): A high-throughput sequencing method for analyzing multiple genes involved in iron metabolism (e.g., TFR2, SLC40A1).

Identifies rare genetic causes of iron deficiency, such as TMPRSS6 gene defects, which disrupt hepcidin regulation.

Proteomics: Analyzes proteins involved in iron metabolism, such as ferritin, transferrin, and hepcidin. Mass spectrometry is used for accurate quantitative determination of these proteins.

MicroRNA (miRNA):MicroRNA (miRNA): miRNAs are small non-coding RNA molecules, approximately 20–25 nucleotides long, that regulate gene expression at the post-transcriptional level. They play a crucial role in controlling many biological processes, including iron metabolism. miRNAs bind to target mRNA (usually in the 3' UTR—untranslated region), leading to either degradation of the mRNA or inhibition of its translation into a protein.

Impact on Iron Metabolism: miRNAs regulate the expression of genes that control iron levels in the body, such as hepcidin (a key hormone that regulates intestinal iron absorption and its release from stores).

miRNAs are also involved in the regulation of iron transport proteins (e.g., ferritin and transferrin).

Examples: miRNA-122 is associated with controlling hepcidin expression. Its dysfunction can alter hepcidin levels, impacting iron absorption and distribution in the body. For instance, during inflammation or chronic diseases, reduced miRNA-122 levels may lead to increased hepcidin expression, contributing to anemia of chronic disease.

miRNA-485: also regulates hepcidin levels and other proteins involved in iron metabolism.

Its imbalance has been linked to pathological conditions, such as iron deficiency anemia or iron overload.

Clinical Significance: Studying miRNA expression provides new opportunities for diagnosing and treating diseases associated with iron metabolism:

miRNAs can serve as biomarkers indicating disruptions in iron metabolism.

They are promising therapeutic targets (e.g., using anti-miRNA strategies to adjust their levels).

CRISPR-based Methods: Used to develop functional models for studying IDA pathophysiology.Analyzes the functional effects of mutations in key genes.

Epigenetic Studies: Investigates DNA methylation and histone modifications that may influence the expression of iron metabolism genes.

Modern Trends: Integration of Artificial Intelligence: **For interpreting laboratory data.**

Comprehensive Approach: Combining clinical assessment, traditional, and molecular methods to ensure accurate diagnostics.

Modern methods for diagnosing IDA significantly enhance detection accuracy and enable the identification of not only the presence of iron deficiency but also its underlying causes. The introduction of molecular methods and new biomarkers ensures a more personalized approach to the diagnosis and treatment of patients.

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