ADVANCES IN BIOMEDICINE FOR THE EARLY DIAGNOSIS OF DISEASES

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Kelly Cruz[1], Larisse Maria de Aguiar Lima[2], Claudiane de Lima Braz[3] and Bruna da Silva Lopes[4]

Abstract

Early diagnosis is one of the most relevant pillars for reducing morbidity and mortality associated with chronic and infectious diseases. Therefore, this chapter aims to analyze the advances of biomedicine in the early diagnosis of diseases, considering technological and biomolecular innovations applied to clinical practice. The methodology was based on a narrative review of scientific literature, including classical and contemporary studies by authors such as Francis Collins, Eric Topol, Leroy Hood, and Harold Varmus, published in international scientific databases. The results indicate that the use of molecular biomarkers, genomic testing, artificial intelligence, and high-precision imaging technologies has significantly increased the ability to identify pathological changes before the clinical manifestation of symptoms. These advances allow faster, more personalized, and more effective interventions, especially in diseases such as cancer, diabetes, and cardiovascular disorders. It is concluded that the integration of biotechnology, genetics, and computational analysis is transforming early diagnosis into an essential tool of precision medicine, promoting greater therapeutic efficiency, reduced healthcare costs, and improved quality of life for patients.

Keywords: Biomedicine. Biomarkers. Early diagnosis. Precision medicine.

[1] Graduate in Biological Sciences – FAVENI
Postgraduate in Clinical Analyses – FAVENI
E-mail: kcfisica@gmail.com
[2] Undergraduate student in Biomedicine
Centro Universitário Unifanor Wyden
E-mail: Larisselari809@gmail.com
[3] Graduate in Biomedicine – Uniasselvi
E-mail: Annelimabraz@gmail.com
[4] Undergraduate student in Biomedicine
Unama
E-mail: brunalopes1929@gmail.com

INTRODUCTION

Advances in biomedicine have brought about significant transformations in health systems, especially with regard to the early detection of diseases. The incorporation of technologies such as genomics, proteomics, molecular biomarkers, artificial intelligence, and high-precision imaging examinations has expanded the capacity to identify pathophysiological changes at early stages, often before the onset of clinical symptoms. This progress represents a milestone for contemporary medicine, as it enables faster, more personalized, and more effective interventions, directly impacting patients' survival and quality of life.

Despite these advancements, challenges remain concerning accessibility, standardization of diagnostic methods, and the integration of these technologies into everyday clinical practice. Accordingly, the research problem is delimited by the following question: in what ways have advances in biomedicine contributed to the early diagnosis of diseases, and what are their impacts on prevention, treatment, and clinical outcomes?

The general objective of this chapter is to analyze the main advances in biomedicine applied to the early diagnosis of diseases. The specific objectives are to: (a) identify the principal biomedical technologies used in early detection; (b) discuss the role of biomarkers and genomic analyses in this process; (c) assess the clinical benefits of early diagnosis; and (d) reflect on the challenges and future prospects of diagnostic biomedicine.

The justification for this study is grounded in the growing incidence of chronic, degenerative, and infectious diseases, which require more efficient detection and intervention strategies. Late diagnosis remains one of the main causes of clinical deterioration and increased healthcare costs, making it essential to expand knowledge about biomedical tools capable of identifying pathological processes at an early stage.

From a theoretical perspective, modern biomedicine is supported by the principles of precision medicine, proposed by authors such as Francis Collins and Leroy Hood, who advocate the use of genetic,

molecular, and environmental information to individualize healthcare. Furthermore, studies by Eric Topol and Harold Varmus highlight the role of biotechnology, artificial intelligence, and molecular biology in constructing a more sensitive, predictive, and preventive diagnostic model. These theoretical contributions underpin the relevance of biomedical advances as central elements in the evolution of early diagnosis and in the consolidation of a more effective and personalized medicine.

METHODOLOGY

TYPE AND DESIGN OF THE RESEARCH

This chapter was developed based on qualitative research, with an exploratory and descriptive character, grounded in a narrative review of the scientific literature. This approach makes it possible to analyze, interpret, and integrate knowledge produced by different areas of biomedicine, fostering a broad and critical understanding of advances in the early diagnosis of diseases.

SEARCH STRATEGY AND STUDY SELECTION

The literature search was conducted in recognized scientific databases, such as PubMed, Scopus, Web of Science, and SciELO, using descriptors in Portuguese and English, including “biomedicine,” “early diagnosis,” “biomarkers,” “precision medicine,” and “genomics.” The terms were combined using Boolean operators (AND, OR) to broaden and refine the results.

We included scientific articles, systematic reviews, guidelines, and books published preferably between 2010 and 2024 that addressed biomedical applications in the early diagnosis of diseases. Duplicated studies, texts without peer review, or works not directly related to the theme were excluded.

INSTRUMENTS AND DATA COLLECTION PROCEDURES

The main data collection instrument was a bibliographic analysis protocol designed to record information such as authors, year of publication, type of biomedical technology, clinical area of

application, and main results. The selected texts were read in full, allowing the extraction of data relevant to the construction of analytical categories.

STUDY SAMPLE

The sample consisted of national and international scientific publications addressing the use of biomedical tools in early diagnosis, including studies on molecular biomarkers, genetic tests, advanced imaging techniques, and applications of artificial intelligence. This diversity ensures a comprehensive view of the area's technological and clinical advances.

DATA ANALYSIS TECHNIQUE

Data were analyzed using thematic content analysis, as proposed by Bardin, enabling the organization of findings into categories such as: diagnostic technologies, clinical impact, benefits of early diagnosis, and implementation challenges. This technique enables the systematic interpretation of results and their correlation with the theoretical frameworks of biomedicine.

METHODOLOGICAL FOUNDATION

The choice of a narrative review is justified by the need to integrate different scientific perspectives on early diagnosis, as advocated by authors such as Collins, Hood, and Topol, who emphasize the interdisciplinary nature of modern biomedicine. Thus, the methodology adopted not only describes technological advances but also critically discusses their clinical, social, and scientific impacts.

RESULTS AND DISCUSSION

Analysis of the scientific literature revealed that advances in biomedicine have decisively impacted the early diagnosis of diseases, promoting a transition from the traditional model—centered on clinical manifestation—to a predictive, preventive, and personalized model. Among the main findings, the

use of molecular biomarkers, genomic analyses, high-resolution imaging technologies, and the application of artificial intelligence in clinical decision support stands out.

Biomarkers have become fundamental tools for the early detection of pathological processes. Studies by Varmus and Hood demonstrate that molecular alterations can be identified long before the emergence of clinical signs, enabling faster and more targeted interventions. In cancer, for example, the identification of specific mutations through genetic tests and liquid biopsies has made it possible to diagnose at early stages, significantly increasing survival rates.

Another relevant finding concerns the advancement of imaging technologies, such as functional magnetic resonance imaging, positron emission tomography (PET), and hybrid examinations, which allow the visualization of metabolic and structural alterations in early phases of diseases. According to Topol, integrating these technologies with artificial intelligence algorithms has increased diagnostic accuracy, reduced human errors, and improved the efficiency of health systems.

The analysis of studies also highlights the central role of genomics and precision medicine. Collins emphasizes that interpreting the human genome makes it possible to identify genetic predispositions to various pathologies, enabling personalized preventive strategies. In this way, early diagnosis ceases to be merely reactive and becomes predictive, based on individual risk.

Taken together, the results indicate that modern biomedicine promotes a paradigmatic shift in the way diseases are diagnosed. However, the literature also points to challenges related to accessibility, the cost of technologies, and the need for professional training—factors that still limit the full incorporation of these innovations into clinical practice.

CONCLUSION

This chapter aimed to analyze advances in biomedicine for the early diagnosis of diseases, considering the contributions of biomolecular, genomic, computational, and imaging technologies to contemporary clinical practice. Based on a review of the scientific literature, it was possible to understand

how biomedicine has reshaped the diagnostic process, shifting it from a predominantly reactive model to a predictive, preventive, and personalized paradigm.

The main results showed that the use of molecular biomarkers, genetic tests, liquid biopsies, high-precision imaging examinations, and artificial intelligence systems has significantly expanded the capacity for early detection of various diseases—especially cancers, cardiovascular diseases, metabolic disorders, and neurodegenerative conditions. These resources make it possible to identify cellular, genetic, and metabolic alterations before the appearance of clinical manifestations, which favors therapeutic interventions that are faster, more effective, and less invasive. In addition, the incorporation of precision medicine allows treatment to be tailored to the individual characteristics of each patient, increasing the likelihood of therapeutic success and reducing adverse effects.

In scientific and social terms, this study contributes to strengthening the understanding of biomedicine as a strategic area for the sustainability of health systems. By promoting earlier diagnoses, biomedicine not only improves clinical outcomes but also contributes to reducing hospital costs, rationalizing the use of therapeutic resources, and enabling more efficient planning of public health policies. Thus, early diagnosis emerges not only as a clinical tool but also as an instrument for the management and promotion of collective health.

Nevertheless, the literature also highlights important challenges, such as the high cost of some technologies, unequal access to advanced examinations, the need for professional training, and ethical issues related to the use of genetic data and artificial intelligence. These factors indicate that, although promising, technological progress must be accompanied by regulatory policies, investments in infrastructure, and strategies to broaden access to health services.

Finally, it is suggested that future research deepen the evaluation of the clinical and economic effectiveness of biomedical technologies for early diagnosis in different population contexts, as well as investigate models for integrating technological innovation with primary health care. Studies are also

recommended on the ethical, legal, and social impacts of precision medicine, to ensure that the benefits of biomedicine are widely distributed and sustainable in the long term.

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