MEDS Basic Medicine (2024)
Clausius Scientific Press, Canada

DOI: 10.23977/medbm.2024.020213
ISSN 2616-2091 Vol. 2 Num. 2

High-throughput screening of medical disease biomarkers and the evaluation of their value in early diagnosis

Yuheng Zhang[1,a,*,#], Jiaqi Ren[1,b,#], Runxiu Chen[1,c], Han Wang[1.d]

[1]College of Medical, Shihezi University, Shihezi, Xinjiang, China
[a]2498294312@qq.com, [b]1911513507@qq.com, [c]chen013001300130@yeah.net, [d]1832302523@qq.com
[*]Corresponding author
[#]These authors contributed equally to this work.

Keywords: Biomarkers, high-throughput screening, early diagnosis, medical diseases

Abstract: Internal medicine diseases pose a major threat to global health, and early diagnosis is the key to improving treatment effectiveness. This study used high-throughput screening techniques to successfully identify potential biomarkers associated with the occurrence and development of internal diseases. Early detection of these biomarkers offers new possibilities for disease diagnosis, and their expression levels are closely associated with the disease process. The study also analyzes the advantages and limitations of high-throughput screening technology and proposes corresponding improvement strategies. Despite progress in the screening and evaluation of biomarkers, challenges are needed to improve technical sensitivity, specificity and translation of clinical applications. Future studies are needed to further explore the clinical application potential of these biomarkers and to develop more precise and efficient diagnostic methods.

1. Introduction

Biomarkers are a class of indicators that can reflect normal biological processes, pathological processes, or produce a response to therapeutic interventions. Biomarkers play a crucial role in the diagnosis and management of medical diseases. They can help physicians make an accurate diagnosis in the early stages of the disease, assess disease severity, monitor treatment efficacy, and predict disease recurrence and progression.[1]

1.1 Theoretical basis of biomarkers

The theoretical basis of biomarkers is rooted in molecular biology, genetics, and pathophysiology. For example, tumor biomarkers may include specific proteins expressed by tumor cells, circulating tumor DNA (ctDNA), or tiny RNA (miRNA). These molecules can be tested by blood or other biological samples, thus providing direct evidence for the presence of a tumor. Moreover, biomarkers can reflect the subtype of disease, the genetic background of patients, and sensitivity to specific treatments.

1.2 Progress in high-throughput screening technology

High-throughput screening technology allows scientists to test thousands of biomarker candidates in a single experiment. GeneChiarray technology assess gene expression patterns by analyzing DNA microarrays, help to identify gene expression changes associated with a specific disease. Mass spectrometry enables the accurate identification and quantification of proteins and peptides in biological samples, which is crucial for protein biomarker discovery. Flow cytometry uses a multiparametric analysis of individual cells to identify cell surface markers or biomarkers within the cell.[2]

1.3 Theoretical development and application and practice of early diagnosis technology

Theoretical developments in early diagnostic techniques have focused on improving the sensitivity and specificity of testing, as well as reducing false positive and false negative results. For example, by using advanced bioinformatics tools and algorithms, researchers can screen out the most promising biomarkers from complex biological data. In applied practice, researchers are faced with how to translate these theories into clinically feasible tests. This includes ensuring test reproducibility, standardization, and clinical validation. Furthermore, one needs to consider the cost-effectiveness ratio of testing, patient acceptance, and integration of the healthcare system.

To achieve early diagnosis, researchers are developing more precise combinations of biomarkers that can improve diagnostic accuracy. For example, by combining multiple biomarkers and clinical information. Moreover, with the development of liquid biopsy techniques, non-invasive early diagnostic tests are becoming possible, which will greatly improve patients' diagnostic experience. High-throughput screening technology was used to identify a range of potential medical disease biomarkers. Through rigorous experimental design and data analysis, we found that those biomarkers were detectable in the early stages of disease and that the expression levels of these biomarkers were strongly associated with disease severity and response to treatment. These findings will provide a rationale for the development of new diagnostic tools and treatments, and ultimately, improve clinical outcomes for patients.[3]

2. Current situation analysis and problem identification

The application of biomarkers and high-throughput screening techniques in the early diagnosis of medical diseases is full of potential, but it also has significant theoretical challenges and practical barriers. First, the understanding of the biological properties and pathological significance of biomarkers is still limited, and deeper basic research is needed to clarify their specific role in the occurrence and development of disease. Second, although high-throughput screening techniques can handle large numbers of samples, the interpretation and validation of their results are often complex and time-consuming, requiring detailed analysis combining bioinformatics tools and statistical methods. Stability and reproducibility of biomarkers are key issues in clinical applications. Sample collection, processing, and storage conditions may significantly affect the activity and concentration of biomarkers, leading to inconsistent results. Therefore, it is essential to establish a standardized sample processing and analysis process. In clinical practice, how to effectively integrate biomarker testing with existing diagnostic processes and treatment strategies is also an important challenge to realize its clinical value.[4]

Many potential biomarkers show promise in laboratory studies, but only a few are able to use in clinical practice as standard tools through rigorous clinical validation and regulatory approval. The high cost, long period, and complexity in this process limit the rapid transformation and application of new biomarkers. Therefore, future research requires more efforts to improve the scientific

validation, technology optimization and clinical application of biomarkers, and some challenges and problems remain to be addressed.

2.1 Limitations of the high-throughput screening techniques

Despite the important role of high-throughput screening techniques in the identification of biomarkers, these techniques have several limitations. First, high-throughput screening often produces large amounts of data, which requires sophisticated bioinformatics tools for analysis and interpretation. Furthermore, these techniques may lack sufficient specificity to cause an increase in false-positive results. Second, many candidates for biomarkers showed potential in the initial screening, but failed to be confirmed in subsequent validation studies. This may be due to an insufficient number of samples, experimental design defects, or biological variability. Finally, high-throughput screening techniques are relatively costly, which limits their application in resource-limited settings.

2.2 Challenges of early diagnostic techniques

Challenges for early diagnostic techniques include improving the sensitivity and specificity of testing, simplifying the test process, reducing the costs, and ensuring the reproducibility and accuracy of test results. For example, many existing biomarker detection methods require sophisticated laboratory equipment and specialized technicians, which limits their application in primary care settings. Furthermore, the development of early diagnostic tests requires a rigorous clinical validation process, which is often time-consuming and costly. Furthermore, even if a biomarker shows potential in research, its translation into clinical practice needs to address regulatory, ethical, and legal issues.[5]

2.3 Gap between existing theoretical research and clinical practice

The gap between theoretical research and clinical practice is one of the main obstacles to achieving the clinical application of biomarkers. Theoretically, many biomarkers show strong predictive power and disease diagnostic potential, but in clinical practice,

3. Theoretical challenges and practical obstacles

In the field of early diagnosis of internal medicine diseases, the study of biomarkers and the application of high-throughput screening technology are two key development directions. They theoretically offer the possibility of early detection of the disease, and in practice they provide a means of efficiently screening the relevant biomarkers. However, in the transformation process from theory to practice, we have encountered a series of challenges and obstacles. Theoretical challenges and practical barriers are inevitable problems in biomarker research and application. It is necessary to explore the main problems encountered in the development of biomarker screening and early diagnosis technology.

3.1 Theoretical challenges in biomarker screening

Theoretical challenges mainly include poor understanding of the biological properties of biomarkers and how to screen biomarkers of clinical value from a large number of candidate markers. Moreover, the heterogeneity of the disease leads to the complexity of the biomarker expression patterns, making it difficult for a single marker to apply to all patients. Another

challenge is how to understand and apply biomarker changes in disease progression and how these changes influence diagnosis and treatment decisions.[6]

3.2 Practical difficulties of early diagnosis

In practice, challenges facing early diagnosis include difficulties in sample collection, problems with the stability of biomarkers, and the sensitivity and specificity of detection methods. For example, for the early diagnosis of tumors, obtaining sufficient tumor tissue for biomarker analysis can be very difficult. Moreover, the preservation and treatment conditions of the biological samples may affect the stability and reliability of the biomarkers. The sensitivity and specificity of the detection methods also need to be further improved to reduce misdiagnosis and missed diagnosis.

3.3 Borders to clinical application

Transof biomarkers from laboratory studies to clinical applications faces multiple hurdles. First is the cost issue, and many high-throughput screening techniques and biomarker detection methods are costly, limiting their widespread use in the clinic. Second, regulatory barriers, new biomarkers and testing methods need to go through a rigorous approval process, which is not only time consuming but may limit the rapid application of innovative technologies. Moreover, the acceptance of new biomarkers by physicians and patients is also one of the challenges in implementing clinical applications. Increasing awareness and trust in biomarkers among healthcare professionals through education and training.[7]

4. Countermeasures research and theoretical innovation

In the field of early diagnosis of internal medicine diseases, biomarkers and high-throughput screening techniques play a key role. Biomarkers help identify the presence and progression of disease, while high-throughput screening techniques are able to rapidly identify these markers. Still, these areas face challenges: sample processing and storage conditions may affect the biomarker stability and require complex data analysis. Moreover, in clinical application, how to integrate these markers into the existing diagnostic process and how to overcome the high cost and long cycle of regulatory approval are all issues to be addressed. Future research should focus on improving the scientific validation, technology optimization, and clinical application of biomarkers to ensure that these tools can be effectively translated into clinical practice to improve disease diagnosis and patient care.

4.1 Improvement strategies for high-throughput screening techniques

To improve the efficiency and accuracy of high-throughput screening techniques, researchers can employ multiple strategies. First, through algorithm optimization and machine learning techniques, the accuracy of data analysis can be improved to reducing false positive and false negative results. Secondly, the development of novel experimental design and statistical methods can enhance the screening capacity of biomarker candidates. Furthermore, utilizing multi-omics data integration strategies, such as combining genomics, proteomics, and metabolomics data, can provide a more comprehensive biomarker analysis.[8]

4.2 Theoretical method for the value assessment of biomarkers

Value assessment of biomarkers requires a systematic set of theoretical approaches. Researchers

can employ statistical learning techniques such as logistic regression, decision trees, and random forests to evaluate the predictive power of biomarkers. At the same time, the use of bioinformatics tools to analyze the biological functions and mechanism of action of biomarkers can deepen the understanding of their potential for clinical application. Moreover, the establishment of a clinical validation framework for biomarkers, including prospective cohort studies and intervention studies, is key to evaluating their practical application value.

4.3 Optimization of early diagnosis techniques

Optimizing the early diagnosis technology needs to start from improving the detection sensitivity and specificity. Researchers can explore new biological detection methods, such as nanotechnology, microfluidic chips and CRISPR gene editing, that provide higher sensitivity detection. At the same time, the development of multi-marker combination detection method can improve the accuracy of diagnosis. Furthermore, early diagnosis techniques can be verified and optimized through large-scale collection and analysis of clinical samples.[9]

4.4 Promotion measures for clinical application

Promote the translation of biomarkers in clinical application. First, strengthening communication with regulators could speed up the approval process for new biomarkers. Secondly, the commercialization and clinical application of biomarker detection technology can be promoted through the cooperation with enterprises and medical institutions. Furthermore, increased awareness of biomarkers among medical professionals and patients can be achieved through education and training. Finally, the establishment of standard operation procedures and quality control system for biomarker testing can ensure the reliability and consistency of testing results.

5. Summary and suggestions

This study has made remarkable progress in the identification and evaluation of medical disease biomarkers through high-throughput screening techniques. Research reveals the potential of specific biomarkers in early diagnosis of disease and provides a rationale for future diagnostic tool development. Studies have successfully screened out multiple potential biomarkers associated with medical diseases, and the early detection of these markers may help in the timely diagnosis of the disease. Meanwhile, studies also found that the expression levels of these biomarkers were strongly associated with disease severity and response to treatment. These findings not only enrich the biomarker repertoire for medical diseases, but also offer the possibility of early intervention in diseases. Moreover, the methodology of this study provides a reference for biomarker studies in other complex diseases and has important theoretical and practical significance. Future studies should continue to explore the clinical application potential of these biomarkers and conduct large-scale clinical trials to validate their diagnostic efficacy. At the same time, the optimization of biomarker detection technology and cost-effectiveness analysis should be enhanced to facilitate its application in clinical practice.

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