

Next-Generation Sequencing in Disease Diagnosis

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ABOVE THE STUDY

Next-Generation Sequencing (NGS) has fundamentally transformed disease diagnosis by enabling comprehensive, high-throughput analysis of genetic material at an unprecedented scale and resolution. In my view, NGS represents one of the most significant technological shifts in modern clinical medicine, moving diagnostics from targeted, hypothesis-driven testing toward unbiased, genome-wide exploration. This transition has not only improved diagnostic accuracy but also expanded our understanding of disease mechanisms, particularly in complex and heterogeneous conditions.

At its core, NGS allows simultaneous sequencing of millions of fragments, making it possible to analyze entire genomes, exomes, or targeted gene panels in a single experiment. This capability has dramatically accelerated the identification of genetic variants associated with disease. In clinical diagnostics, NGS is now widely used to detect inherited disorders, characterize cancers, and identify infectious agents. Its ability to uncover rare or unexpected mutations has been especially valuable in cases where traditional diagnostic methods fail to provide answers.

One of the most impactful applications of NGS is in rare genetic diseases. Many of these conditions are caused by mutations in a wide range of genes, often with overlapping clinical symptoms. Traditional single-gene testing approaches are inefficient and time-consuming in such scenarios. NGS enables simultaneous screening of multiple genes, significantly increasing diagnostic yield and reducing the “diagnostic odyssey” experienced by many patients. In my opinion, this represents a major step forward in equitable healthcare, as it provides answers to patients who previously remained undiagnosed for years.

In oncology, NGS has revolutionized the concept of precision medicine. Tumors are genetically heterogeneous, and their behavior is driven by complex mutational landscapes. NGS allows comprehensive profiling of tumor genomes, identifying driver mutations, resistance mechanisms, and actionable therapeutic targets. This information enables clinicians to select targeted therapies tailored to the molecular characteristics of

each patient’s tumor. Additionally, serial sequencing can monitor tumor evolution and detect emerging resistance mutations, supporting adaptive treatment strategies.

NGS has also become an essential tool in infectious disease diagnostics. Metagenomic sequencing can identify pathogens directly from clinical samples without prior knowledge of the causative organism. This is particularly useful in cases of atypical infections, outbreaks, or when conventional microbiological methods fail. During recent global health crises, NGS played a crucial role in identifying novel pathogens and tracking viral mutations in real time. In my view, this real-time surveillance capability is one of the most powerful aspects of NGS in public health.

Despite its advantages, several challenges limit the widespread clinical implementation of NGS. One of the primary issues is data interpretation. Sequencing generates vast amounts of information, including numerous variants of uncertain significance. Distinguishing pathogenic mutations from benign polymorphisms requires robust bioinformatics tools and well-curated reference databases. This interpretative complexity can delay clinical decision-making and requires specialized expertise.

Another important challenge is cost and accessibility. Although sequencing costs have decreased significantly over the past decade, NGS remains expensive for routine use in many healthcare systems, particularly in low- and middle-income countries. Infrastructure requirements, including sequencing platforms, computational resources, and trained personnel, further limit accessibility. In my opinion, addressing these disparities is essential to ensure that the benefits of genomic medicine are globally equitable.

Ethical and legal considerations also play an important role in NGS-based diagnostics. Issues such as incidental findings, genetic privacy, data ownership, and informed consent are increasingly relevant as whole-genome sequencing becomes more common. Patients may receive information about unrelated genetic risks, raising complex questions about disclosure and psychological impact. Clear guidelines and counseling

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frameworks are necessary to manage these challenges responsibly.

Looking forward, integration of NGS with other omics technologies such as transcriptomics, proteomics, and metabolomics will likely enhance diagnostic precision further. Additionally, artificial intelligence and machine learning are expected to play a major role in interpreting complex genomic datasets and predicting disease outcomes. In my view, the convergence of these technologies will define the next era of precision diagnostics.

In conclusion, next-generation sequencing has reshaped disease diagnosis by enabling comprehensive, high-resolution genetic analysis across a wide range of clinical applications. While challenges remain in interpretation, cost, and ethical management, its impact on rare disease diagnosis, oncology, and infectious disease control is undeniable. I believe that as technology continues to advance and become more accessible, NGS will form the backbone of future diagnostic medicine, enabling truly personalized and predictive healthcare.