



Next-generation sequencing: Accelerating gene discovery

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INTRODUCTION

Next-Generation Sequencing (NGS) has emerged as a transformative technology in the field of genomics, revolutionizing the way researchers study and understand the intricacies of the genetic code. Since its inception, NGS has rapidly evolved, offering unprecedented speed, scalability, and cost-effectiveness compared to traditional Sanger sequencing methods. This technological revolution has accelerated the pace of gene discovery and propelled advancements in diverse fields, from basic research to clinical diagnostics and beyond.

At the heart of NGS lies the ability to sequence DNA molecules in a massively parallel fashion, enabling the simultaneous analysis of millions to billions of DNA fragments. This high-throughput approach has dramatically increased sequencing throughput while reducing the time and resources required generating large-scale genomic data. As a result, NGS has democratized access to genomic information, empowering researchers and clinicians worldwide to unravel the complexities of the genome with unprecedented efficiency.

DESCRIPTION

The advent of NGS has led to a paradigm shift in genomics research, fueling a surge in Genome-Wide Association Studies (GWAS), transcriptomics, epigenomics, metagenomics, and other-omics disciplines. By generating vast amounts of sequencing data, NGS has facilitated genome-wide surveys of genetic variation, gene expression, DNA methylation, chromatin accessibility, and microbial diversity, among other molecular features. These comprehensive datasets have provided invaluable insights into the genetic basis of diseases, developmental processes, evolutionary relationships, and environmental interactions.

NGS has played a pivotal role in elucidating the genetic architecture of complex traits and diseases, from common disorders like diabetes and cardiovascular disease to rare genetic syndromes and cancers. By sequencing the genomes of thousands to millions of individuals, researchers have identified genetic variants associated with disease susceptibility, drug response, and clinical outcomes. These findings have not only deepened our understanding of disease mechanisms but also paved the way for precision medicine approaches tailored to individual patients' genetic profiles.

Moreover, NGS has enabled the discovery of novel genes, regulatory elements, and genetic pathways underlying diverse biological phenomena. Through whole-genome sequencing, exome sequencing, and targeted gene panels, researchers have uncovered rare and de novo mutations responsible for Mendelian disorders, intellectual disabilities, and developmental disorders. Additionally, NGS-based approaches have revealed the regulatory mechanisms governing gene expression, alternative splicing, and post-transcriptional modifications,

providing insights into the complexity of gene regulation and its dysregulation in disease states.

In the realm of infectious diseases, NGS has revolutionized our ability to track and monitor microbial pathogens, detect antimicrobial resistance, and investigate disease outbreaks. By sequencing the genomes of bacteria, viruses, fungi, and parasites, scientists can infer transmission routes, identify virulence factors, and characterize the genomic diversity of pathogenic strains. This genomic surveillance approach has proven invaluable in guiding public health interventions, designing effective vaccines, and mitigating the spread of infectious agents.

Furthermore, NGS has opened new frontiers in personalized medicine, offering unprecedented opportunities for diagnosing, prognosing, and treating a wide range of medical conditions. By sequencing patients' genomes, researchers and clinicians can identify disease-causing mutations, assess disease risk, and tailor therapeutic interventions based on individual genetic profiles. In oncology, NGS-based tumor profiling has become standard practice for guiding precision oncology treatments, including targeted therapies, immunotherapies, and clinical trial enrollment.

The widespread adoption of NGS in research, clinical, and commercial settings has been facilitated by ongoing advancements in sequencing technologies, bioinformatics algorithms, and data analysis pipelines. Continuous improvements in sequencing platforms, such as Illumina, Pacific Biosciences, and Oxford Nanopore Technologies, have driven down the cost per base pair and increased the throughput and accuracy of sequencing experiments. Concurrently, bioinformatics tools for read alignment, variant calling, and functional annotation have become increasingly sophisticated, enabling researchers to extract meaningful insights from complex genomic datasets.

Despite its transformative potential, NGS still faces several technical and computational challenges that must be addressed to fully realize its promise. Issues such as sequencing errors, read mapping biases, and data storage requirements pose significant hurdles for large-scale genomic studies. Moreover, the interpretation of genomic variants, especially in non-coding regions and complex genetic diseases, remains a formidable task that requires interdisciplinary expertise and collaborative efforts.

Looking ahead, the future of NGS holds promise for further innovation and impact across diverse domains of science and medicine. Emerging technologies, such as single-cell sequencing, long-read sequencing, and spatial transcriptomics, are expanding the scope of genomic research and enabling new discoveries at the cellular and spatial resolution. Additionally, advances in machine learning, artificial intelligence, and cloud computing are poised to enhance the scalability, efficiency, and reproducibility of NGS data analysis workflows.

CONCLUSION

Next-generation sequencing has emerged as a game-changing technology in genomics, catalyzing breakthroughs in gene discovery, disease research, and personalized medicine. By enabling high-throughput, cost-effective analysis of DNA molecules, NGS has democratized access to genomic information and empowered researchers to explore the complexities of the genome with unprecedented depth and breadth. As NGS continues to evolve and mature, it holds the potential to drive further innovation and transform our understanding of the genetic basis of life and disease.