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ISSN: 2161-1041
Hereditary Genetics: Current Research

Commentary

Gene Sequencing: Current Research Driving Genomic Discovery and Precision Medicine

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DESCRIPTION

Gene sequencing has revolutionized modern genetics by enabling scientists to determine the precise order of nucleotides within Deoxyribonucleic Acid (DNA). This technology has become fundamental to biomedical research, clinical diagnostics, evolutionary biology and precision medicine. By identifying genetic variations associated with inherited disorders, infectious diseases and complex traits, gene sequencing has transformed the understanding of human health and disease. Current research continues to improve sequencing accuracy, speed, affordability and data interpretation, making genomic analysis increasingly accessible for both research and clinical applications.

The principle of gene sequencing involves determining the arrangement of the four nucleotide bases adenine, thymine, cytosine and guanine that constitute DNA molecules. Genetic variations, including single nucleotide variants, insertions, deletions, copy number variations and structural rearrangements, can influence gene function and contribute to disease susceptibility. Identifying these variations enables researchers and clinicians to establish molecular diagnoses, investigate disease mechanisms and develop personalized therapeutic approaches.

The introduction of Next-Generation Sequencing (NGS) has dramatically accelerated genomic research by allowing millions of DNA fragments to be sequenced simultaneously. Compared with conventional Sanger sequencing, NGS provides significantly greater throughput, improved sensitivity and reduced costs. Clinical laboratories now routinely employ targeted gene panels, Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS) for diagnosing rare genetic disorders, hereditary cancers, neurological diseases and cardiovascular conditions. Current research continues to optimize these sequencing platforms for enhanced clinical utility.

Whole Genome Sequencing provides comprehensive analysis of both coding and non-coding regions of the genome, enabling detection of nearly all classes of genetic variation. In contrast, Whole Exome Sequencing focuses exclusively on protein-coding regions, which comprise approximately one to two percent of the human genome but harbor a substantial proportion of disease-causing mutations. Depending on the clinical indication, researchers select the most appropriate sequencing strategy to maximize diagnostic efficiency while minimizing unnecessary data generation.

Long-read sequencing technologies have emerged as a major advancement in genomic research. Unlike short-read sequencing, which analyzes relatively small DNA fragments, long-read sequencing generates continuous reads spanning thousands of nucleotides. This capability improves the identification of complex structural variants, repetitive genomic regions and chromosomal rearrangements that are often difficult to detect using conventional sequencing methods. Current studies demonstrate that long-read sequencing significantly enhances diagnostic accuracy for numerous inherited neurological and developmental disorders.

Single-cell sequencing has expanded genomic research by enabling the analysis of individual cells rather than bulk tissue samples. This technology reveals cellular heterogeneity that cannot be detected using conventional sequencing approaches. In oncology, single-cell sequencing helps identify rare cancer cell populations responsible for metastasis and treatment resistance.

Metagenomic sequencing has transformed infectious disease research by allowing comprehensive analysis of microbial communities without requiring traditional culture methods. Clinical applications include rapid identification of bacterial, viral, fungal and parasitic pathogens from patient samples. During infectious disease outbreaks, metagenomic sequencing facilitates pathogen surveillance, transmission tracking and the identification of emerging variants.

In conclusion, gene sequencing has become one of the most transformative technologies in modern genetics and precision medicine. Current research continues to expand its applications in hereditary diseases, cancer genomics, infectious disease

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Received: 23-May-2026, Manuscript No. HGCR-26-32040; **Editor assigned:** 25-May-2026, PreQC No. HGCR-26-32040 (PQ); **Reviewed:** 09-Jun-2026, QC No. HGCR-26-32040; **Revised:** 16- Jun -2026, Manuscript No. HGCR-26-32040 (R); **Published:** 23-Jun-2026, DOI: 10.35248/2161-1041.26.15.311

Citation: Novak L. (2026). Gene Sequencing: Current Research Driving Genomic Discovery and Precision Medicine. Hereditary Genet. 15:311.

surveillance and personalized healthcare. Ongoing technological innovations, coupled with advances in computational biology and artificial intelligence, are expected to further improve diagnostic accuracy, therapeutic decision-making and the understanding of human genetic diversity.