



The Role of DNA Sequencing in Modern Biological Research

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Abstract

DNA sequencing is a fundamental technology in modern biological research because it enables researchers to determine the nucleotide composition of DNA and investigate genetic variation, gene function, evolution, and disease mechanisms. Advances from first-generation Sanger sequencing to next-generation sequencing and long-read platforms have greatly increased sequencing capacity while reducing the time and cost required to generate genomic information. Sequencing is now applied to whole genomes, exomes, transcriptomes, microbial communities, and single cells. It supports fields ranging from molecular biology and evolutionary research to clinical genomics, agriculture, microbiology, and precision medicine. This paper examines nine major applications of DNA sequencing, including genome analysis, genetic variation, disease research, transcriptomics, microbial genomics, evolutionary biology, biodiversity, agricultural research, and emerging single-cell and long-read approaches.

Keywords

DNA Sequencing; Genomics; Next-Generation Sequencing; Sanger Sequencing; Long-Read Sequencing; Whole-Genome Sequencing; Genetic Variation; Transcriptomics; Bioinformatics; Molecular Biology

Introduction

The ability to determine the order of nucleotides in DNA has transformed biological science. Sequencing provides direct molecular information that can be used to characterize genomes, identify genetic differences, investigate gene function, and reconstruct biological relationships.

Early sequencing technologies established the foundation for modern genomics, while high-throughput sequencing platforms made it possible to analyze millions or billions of DNA fragments in parallel. These developments substantially expanded the scale of biological research.

Modern sequencing is closely connected with bioinformatics. Raw sequence data require computational processing, including quality assessment, alignment or assembly, variant detection, annotation, and statistical interpretation. Consequently, sequencing research combines laboratory methods with computational biology.



This paper examines nine major dimensions of DNA sequencing in modern biological research: sequencing technologies, genome analysis, genetic variation, disease research, transcriptomics, microbial genomics, evolutionary studies, agriculture and biodiversity, and emerging single-cell and long-read applications.

1. Evolution of DNA Sequencing Technologies

Sanger sequencing was a major early method for determining DNA sequences and remains useful for targeted sequencing and validation. Its development established principles that later technologies expanded to much larger scales. Next-generation sequencing introduced massively parallel sequencing, allowing researchers to process many DNA fragments simultaneously. This increased throughput and enabled whole-genome and whole-exome studies at scales that were previously difficult to achieve. Third-generation and long-read technologies can produce much longer DNA reads, helping researchers resolve repetitive genomic regions, structural variants, and complex genome assemblies.

2. Whole-Genome and Whole-Exome Sequencing

Whole-genome sequencing examines DNA across essentially the complete genome, providing information about coding and non-coding regions. Whole-exome sequencing focuses primarily on protein-coding regions, which represent a smaller fraction of the genome but contain many disease-associated variants. These approaches are widely used in human genetics, population studies, disease research, and comparative genomics. Sequencing entire genomes can reveal single-nucleotide variants, insertions and deletions, copy-number changes, and structural variants. The choice between whole-genome, whole-exome, and targeted sequencing depends on the research question, required resolution, available resources, and desired genomic coverage.

3. DNA Sequencing and Genetic Variation

Sequencing allows researchers to identify genetic differences among individuals, populations, and species. Variant analysis can reveal single-nucleotide changes, small insertions and deletions, structural variants, and other forms of genomic diversity. Characterizing genetic variation helps researchers investigate inheritance, population structure, adaptation, and disease susceptibility. Large reference datasets also provide information about how frequently particular variants occur in different populations. Accurate interpretation requires attention to sequencing quality, genomic context, population frequency, and supporting functional or clinical evidence.

4. Applications in Disease and Clinical Biology

DNA sequencing has become an important tool for investigating the genetic basis of disease. Researchers can sequence patients, tumors, pathogens, or experimental models to identify genetic changes associated with disease mechanisms. In clinical genetics, sequencing may assist diagnosis of rare inherited disorders and can help characterize



cancers and other conditions. Tumor sequencing can identify molecular alterations that may influence disease classification or therapeutic decisions. The clinical value of sequencing depends on accurate variant interpretation, appropriate validation, and integration of genomic results with medical and phenotypic information.

5. Sequencing in Transcriptomics and Gene-Expression Research

RNA sequencing, commonly called RNA-seq, uses sequencing technologies to characterize RNA molecules after conversion into sequencing-compatible libraries. It provides information about gene expression, transcript abundance, alternative splicing, and other aspects of RNA biology. RNA sequencing allows researchers to compare gene-expression patterns between tissues, developmental stages, treatments, and disease states. It can also identify previously unrecognized transcripts and help connect genetic variation with molecular phenotypes. Combining genomic and transcriptomic sequencing provides a broader view of how DNA differences influence gene activity.

6. Microbial Genomics and Metagenomics

DNA sequencing has transformed microbiology by enabling researchers to study microbial genomes and communities at unprecedented scale. Whole-genome sequencing can identify genes associated with microbial metabolism, virulence, antimicrobial resistance, and environmental adaptation. Metagenomic sequencing examines DNA recovered from mixed microbial communities without requiring every organism to be cultured individually. This approach has expanded understanding of microbiomes in soil, water, food, animals, and humans. Sequencing therefore provides a powerful way to investigate microbial diversity, community structure, and ecological function.

7. Evolutionary Biology and Biodiversity Research

Comparative DNA sequencing allows researchers to reconstruct evolutionary relationships and investigate how genomes change over time. Sequence differences can be used to estimate relationships among organisms and identify patterns of natural selection, migration, and population history. Sequencing has also improved biodiversity assessment by enabling identification of organisms from environmental samples and degraded biological material. DNA barcoding and environmental DNA approaches can support species detection and ecological monitoring. These applications demonstrate how sequencing connects molecular data with large-scale questions about evolution and ecosystems.

8. Agricultural and Environmental Applications

DNA sequencing is increasingly used in agriculture to characterize crop genomes, identify useful genetic variation, study plant and animal breeding populations, and investigate pathogens. Genomic information can support selection of traits related to yield, disease resistance, stress tolerance, and quality. Environmental sequencing can characterize microbial communities and reveal biological responses to changes in soil,

water, climate, and other ecological conditions. Integration of sequencing with phenotypic measurements and computational analysis can support more precise breeding and environmental research.

9. Single-Cell, Long-Read Sequencing, and Future Directions

Single-cell sequencing allows researchers to study genetic or transcriptomic information at the level of individual cells. This can reveal cellular heterogeneity that is hidden when DNA or RNA is analyzed from a mixture of many cells. Long-read sequencing is particularly valuable for resolving repetitive regions, structural variants, haplotypes, and complex genome assemblies. Improvements in accuracy and throughput continue to expand its applications. Future sequencing research is likely to combine long-read methods, single-cell analysis, spatial biology, multi-omics, artificial intelligence, and increasingly comprehensive reference genomes. These developments will provide more detailed views of biological systems and their variation.

Illustration

The following illustration summarizes a simplified DNA-sequencing research workflow, from collection of a biological DNA sample through sequencing, bioinformatic analysis, and interpretation of biological information.

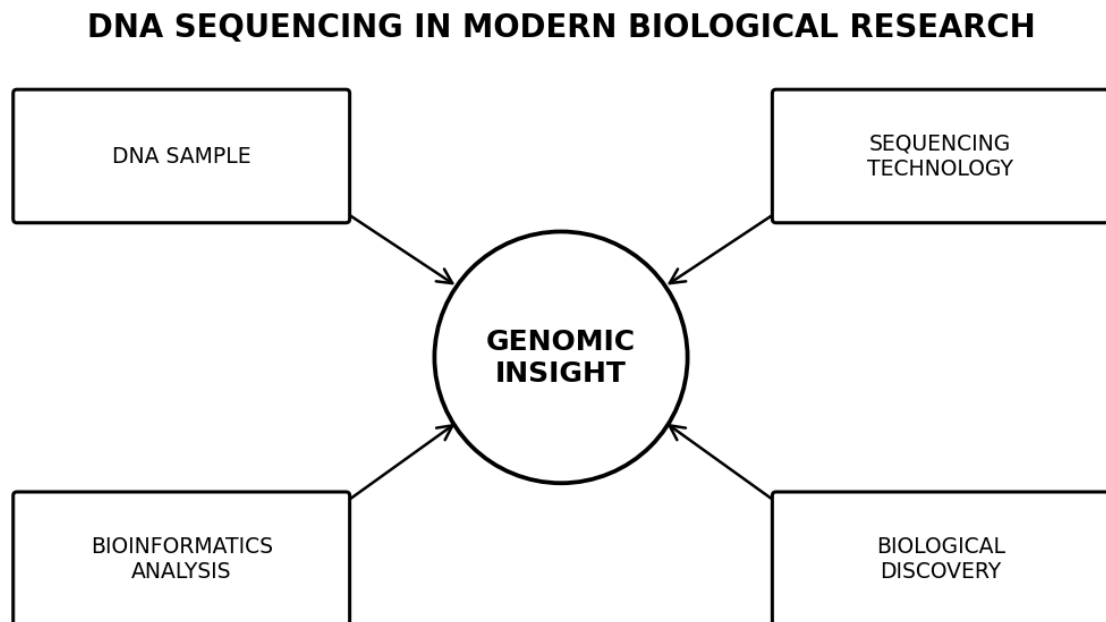


Figure 1. Simplified role of DNA sequencing in modern biological research.



Conclusion

DNA sequencing has become one of the most important technologies in modern biological research. It provides direct information about genetic material and supports investigations ranging from individual genes to complete genomes and complex biological communities.

Advances in high-throughput, long-read, and single-cell sequencing have expanded both the scale and resolution of biological investigation. Sequencing is now central to genetics, molecular biology, microbiology, evolutionary research, agriculture, environmental science, and clinical genomics.

The continuing integration of sequencing with bioinformatics, transcriptomics, spatial analysis, and other omics technologies will further improve understanding of biological systems. As sequencing becomes more comprehensive and accessible, careful data interpretation, quality control, privacy, and responsible use of genomic information will remain important.

References

- Sanger, F., Nicklen, S., & Coulson, A. R. (1977). DNA sequencing with chain-terminating inhibitors. *Proceedings of the National Academy of Sciences*, 74(12), 5463–5467.
- Metzker, M. L. (2010). Sequencing technologies: The next generation. *Nature Reviews Genetics*, 11, 31–46.
- Goodwin, S., McPherson, J. D., & McCombie, W. R. (2016). Coming of age: Ten years of next-generation sequencing technologies. *Nature Reviews Genetics*, 17, 333–351.
- van Dijk, E. L., Auger, H., Jaszczyszyn, Y., & Thermes, C. (2014). Ten years of next-generation sequencing technology. *Trends in Genetics*, 30(9), 418–426.
- Logsdon, G. A., Vollger, M. R., & Eichler, E. E. (2020). Long-read human genome sequencing and its applications. *Nature Reviews Genetics*, 21, 597–614.
- Levy, S. E., & Myers, R. M. (2016). Advancements in next-generation sequencing. *Annual Review of Genomics and Human Genetics*, 17, 95–115.
- Stark, R., Grzelak, M., & Hadfield, J. (2019). RNA sequencing: The teenage years. *Nature Reviews Genetics*, 20, 631–656.