

Next-Generation Sequencing Overview

Next-generation sequencing (NGS) is a powerful and versatile technology that has transformed the field of genomics. With its ability to sequence large amounts of DNA or RNA quickly and cost-effectively, NGS has enabled researchers to gain unprecedented insights into the genetic makeup of organisms and the complex interactions between genes, proteins, and metabolites. From whole genome sequencing to targeted sequencing and beyond, NGS has become an essential tool for researchers in fields ranging from basic science to clinical diagnostics.



Market Size

The NGS market was valued at about \$10 billion in 2022 and is expected to reach \$40 billion by 2030, with an average yearly growth rate of approximately 18.9% from 2023 to 2030.¹ The implementation of precision medicine and demand for point-of-care sequencers are among the factors driving growth in the NGS market.

Key Players^{1,2}

Established and emerging companies have introduced a slew of innovative sequencers over the last few years. This list includes leading instrument providers as well as less familiar but increasingly relevant companies.

- BGI Group
- Complete Genomics
- Element Biosciences
- Illumina
- Oxford Nanopore Technologies
- Pacific Biosciences
- Qiagen
- Roche
- Singular Genomics
- Thermo Fisher Scientific
- Ultima Genomics



NGS Techniques^{3,4}

- **Whole genome sequencing (WGS)** is a comprehensive method that covers the entire DNA sequence of an organism's genome.
- **Whole exome sequencing (WES)** is a method that involves sequencing only the exons from an organism of interest.
- **Targeted DNA sequencing** encompasses a range of methods focused on specific regions of interest in the genome as opposed to WGS or WES.
- **Whole transcriptome sequencing** analyzes the complete set of RNA transcripts in a sample, providing both qualitative and quantitative information.
- **Targeted RNA sequencing** is used to sequence specific transcripts of interest using either capture or amplicon-based approaches.
- **Methylome analysis** is used to study the methylation status of DNA samples and gain insights into gene regulation.
- **Targeted methyl sequencing** allows for sequencing specific regions of interest in the DNA methylome.
- **ChIP-Seq** is used to identify and characterize DNA-binding proteins and histone modifications on a genome-wide scale.

NGS Workflow

NGS workflows generally consist of four main steps: nucleic acid extraction, library preparation, sequencing, and data analysis. During nucleic acid extraction, either DNA or RNA is isolated from a sample. Library preparation then follows, where the nucleic acid material is converted into a format suitable for sequencing. The prepared library is then loaded onto a sequencer. In data analysis, reams of raw sequencing data are taken through a series of steps to process and interpret the genetic information generated by NGS.



Common Applications

- Identifying genes related to cancer⁵
- Development of molecular diagnostics⁶
- Identifying mutation targets for targeted therapy⁷
- Comparing genotypes
- Studying rare disease
- Evaluating epigenetic modifications
- Tracking the spread of pathogens to help manage disease outbreaks

Recent Advances

- Single-cell multiomics focuses on understanding a myriad of interactions by integrating various types of molecular data such as RNA, DNA, chromatin accessibility, and protein expression.⁸⁻¹⁰
- Single-cell sequencing offers several advantages for studying cellular diversity, identifying rare cell types, and detecting somatic genetic variations or mutations that may be masked in bulk sequencing.¹¹



Advantages of NGS

NGS has revolutionized genetic analysis, offering a powerful and efficient alternative to traditional sequencing methods. Its ability to sequence millions of DNA fragments simultaneously, coupled with its cost-effectiveness, speed, and high data resolution, has made it invaluable to researchers. The technology's versatility and accuracy in detecting rare genetic variants have further solidified its position as a leading tool in genetic analysis.

References

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