

“The Future of Genetic Testing: Next Generation Sequencing (NGS)”

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Introduction

Next-Generation Sequencing (NGS)

Definition: A high-throughput method that enables the rapid sequencing of the entire genomes or targeted regions of DNA, allowing for comprehensive analysis of genetic information.

Key Points:

- **High Throughput:** Millions of DNA fragments are sequenced in parallel.
- **Speed:** Significantly faster than traditional sequencing methods.
- **Cost-Effective:** Lower cost per base compared to older technologies.
- **Versatile:** Applicable to various types of genetic studies, including genomics, transcriptomics, and epigenomics.

Research Objective

- To analyze the capabilities and limitations of NGS compared to traditional genetic testing methods.
- To explore the potential applications of NGS in various areas of healthcare (e.g., cancer diagnosis, carrier screening).
- To investigate the ethical considerations surrounding the use of NGS in genetic testing, including informed consent and data privacy.
- To assess the potential economic impact of NGS on healthcare systems.

Research Methodology

Research Design

This qualitative study will use a systematic literature review to explore the future of genetic testing with Next-Generation Sequencing (NGS) technologies.

Data Collection Sources for data include:

- Peer-reviewed journals
- Industry reports
- Government publications
- Online databases (PubMed, Google Scholar, Scopus)

Literature Selection

- Publications from the last ten years
- Articles on NGS technologies and applications in genetic testing
- Studies discussing NGS advantages, challenges, and future prospects

Result & Discussion

Market Growth & Valuation

Global

The next-generation sequencing market is projected to expand from USD 10.47 billion in 2024, at a compound yearly growth rate of 13.43%, to reach USD 19.66 billion by 2029.

Industry leaders are leveraging NGS technology for drug development. A notable collaboration occurred in January 2022 between Illumina Inc. and Nashville Biosciences LLC, a VUMC subsidiary. This multi-year agreement aims to accelerate drug development and establish a leading clinical genomic resource using Illumina's NGS platforms.

Study Period	2019 - 2029
Market Size (2024)	USD 10.47 Billion
Market Size (2029)	USD 19.66 Billion
CAGR (2024 - 2029)	13.43 %
Fastest Growing Market	Asia-Pacific
Largest Market	North America

Next-generation Sequencing (NGS) Market

Market Size in USD Billion
CAGR 13.43%



Result & Discussion

The Future of Genetic Testing: Next-Generation Sequencing Capacities of NGS:

- **Higher Throughput:** Sequences millions of DNA fragments simultaneously.
- **Higher Speed:** Takes hours versus weeks/months for Sanger sequencing.
- **Wide Range of Applications:** Detects various genetic alterations, including single nucleotide and structural variants.
- **Improved Sensitivity and Specificity:** More accurate than Sanger sequencing for small genetic changes.
- **Cost-Effective:** Economical for large-scale studies.

Result & Discussion

Potential Applications in Healthcare:

- **Cancer Diagnosis and Treatment:** Detailed tumor profiling for personalized treatment.
- **Carrier Screening:** Guides reproductive decisions and early interventions.
- **Prenatal Testing:** Noninvasive screening for chromosomal abnormalities.
- **Infectious Disease Diagnostics:** Identifies pathogens, including novel and resistant strains.
- **Rare Genetic Disorders:** Identifies causative mutations for precise diagnosis and treatment.

Ethical Considerations:

- **Informed Consent:** Ensure patients understand the test and its potential findings.
- **Privacy and Security:** Protect genetic data from unauthorized access and misuse.
- **Equity and Access:** Ensure NGS is accessible to diverse populations, minimizing healthcare disparities.

Result & Discussion

Economic Impact on Healthcare:

- **Cost Savings:** Facilitates early diagnosis and targeted treatments, reducing overall costs.
- **Infrastructure Investment:** Requires investment in bioinformatics and trained personnel.
- **Health Economic Evaluations:** Assess the cost-effectiveness of NGS considering direct testing costs and broader economic benefits from improved patient outcomes.

Conclusion

Next-generation sequencing (NGS) holds immense potential for genetic testing, driven by advancements and expanding clinical applications. NGS offers high accuracy, speed, and affordability, revolutionizing healthcare with precise diagnosis, treatment, and disease prevention tailored to individual genetic profiles.

However, these advancements raise ethical and regulatory challenges, including privacy concerns and genetic discrimination. Robust frameworks for informed consent, data security, and genetic counseling are essential. Market leaders like Illumina and Thermo Fisher Scientific drive innovation, enhancing accessibility and affordability. NGS is poised to transform personalized medicine, offering early disease diagnosis and tailored treatments, shaping the future of healthcare.

Recommendations and Suggestions

1. Promote Technological Innovation

- Incentivize R&D in NGS technologies.
- Foster collaborations between academia, industry, and government.

2. Expand Clinical Applications

- Integrate NGS in diagnostics, treatment plans, and preventive care.
- Highlight successful studies and pilot programs.

3. Address Ethical and Privacy Concerns

- Develop policies for ethical concerns and data protection.
- Ensure robust informed consent and genetic counseling protocols.

4. Strengthen Regulatory Frameworks

- Update regulations to match technological advancements.
- Set standards for accuracy, reliability, and utility.

5. Enhance Public and Professional Education

- Train healthcare professionals on NGS interpretation and integration.
- Launch public information campaigns on genetic testing.

Limitation of Study

1. Accuracy and Error Rates

- Sequencing errors and difficulties in detecting some genetic variations affect result accuracy.

2. High Financial Costs and Accessibility

- NGS remains expensive and often not covered by insurance, reducing accessibility in low-resource settings.

3. Ethical and Privacy Concerns

- Handling and storage of genetic data pose privacy risks; obtaining informed consent for NGS is challenging.

4. Regulatory and Standardization Challenges

- Variability in regulatory frameworks and lack of standardization impact reliability and global applicability.

5. Population Diversity

- Genomic research is biased towards populations of European descent, leading to potential health disparities in other ethnic groups.

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