

Safeguarding Africa's Health



Generic Foundational Course

Introduction to Genomic Sequencing and NGS

NGS Academy for the Africa CDC









Introduction to Genomic Sequencing and NGS

back to the table of modules

Module last updated: December 2024

Number of sessions	2–3
Total learning time	6–8 hours
Target audience	All personas - wet laboratory personnel (i.e., scientists, laboratory technicians, etc.), dry laboratory personnel (epidemiologists, bioinformatics scientists, and bioinformaticians), and managerial personnel (i.e., HODs, laboratory managers, policymakers, etc.).
Format	Lectures, videos
Level of the module	Introductory



Contributors

Elizabeth Temiloluwa Akande, Temesgen Endalew, Siddiqah George, Carolina Matos, and Mohammed Ahmed Rameto.



Module description

This module introduces the topic of next-generation sequencing (NGS), a technology that allows for the rapid and accurate sequencing of multiple genes or whole genomes simultaneously. NGS has provided breakthroughs in understanding microbial diversity, evolution, and transmission, which uniquely positions this technology in pathogen surveillance and disease outbreak models. In this module, participants are introduced to the following topics and/or concepts:

- Key concepts in genomic sequencing and its role in genomics
- Traditional sequencing versus next-generation sequencing (NGS)
- The key steps in a typical NGS workflow
- An introduction to different NGS approaches (e.g., whole genome sequencing, targeted enrichment NGS, metagenomics (mNGS), and shotgun metagenomic sequencing)
- The historical development of sequencing technologies
- The fundamental principles and methods of different NGS platforms (e.g., Illumina, Ion Torrent, PacBio, and Oxford Nanopore)
- The advantages and limitations of these various NGS technologies
- Applying key study design principles to NGS

- An overview of NGS data collection and storage
- The basic steps in NGS data analysis
- An introduction to the key bioinformatics tools used in NGS data analysis
- Common file formats used in NGS data (e.g., FASTQ, BAM, and VCF)
- Fundamental principles in sequence alignment and assembly
- The applications of NGS in biomedical research and pathogen genome surveillance

Module learning outcomes

On completion of this module, the participants will have a basic understanding of, or will be able to:

- Explain the fundamental principles of genomic sequencing and its role in genomics
- Compare and contrast traditional versus next-generation sequencing (NGS)
- · List the key steps in the NGS process and understand their purpose
- Describe the advantages and disadvantages of different NGS approaches
- Briefly discuss the history of the development of sequencing technologies
- Differentiate between short- and long-read sequencing technologies
- Compare the different principles and methods utilised by various NGS platforms
- List the advantages and disadvantages of Illumina, Ion Torrent, PacBio, and Oxford Nanopore sequencing platforms
- Apply key study design principles to NGS
- Discuss NGS data collection and storage
- List the basic steps in NGS data analysis
- · Identify the key bioinformatics tools used in NGS data analysis
- Compare and contrast NGS data file formats such as FASTQ, BAM, and VCF
- Discuss the basic concepts in sequence alignment and assembly
- Explain how NGS can be applied in biomedical research and pathogen genome surveillance



Module assessments

Module practical: Not applicable

Module quiz: Assessment questions available on the ASLM platform



Module resources

- <u>Thermo Fisher Scientific Video Next Generation Sequencing Acronyms and Terms</u>
- <u>ASM Training Videos Training in Infectious Disease Applications of NGS</u>
 - The Evolution of NGS Technologies Part 1
 - The Evolution of NGS Technologies Part 2
 - <u>Define Different Sequencing Techniques and Platforms</u>
- NIH | NLM Article Overview of Next Generation Sequencing Technologies
- Thermo Fisher Scientific Video Next Generation Sequencing for Gene Expression Analysis
- <u>CABANA Slides Sequencing technologies</u>
- Illumina: An introduction to Next-Generation Sequencing Technology
- Illumina: NGS Workflow Steps | Illumina sequencing workflow
- Illumina: Differences Between NGS and qPCR
- Illumina: Targeted next-generation sequencing versus qPCR and Sanger sequencing
- Thermo Fisher Scientific Video Ion Torrent Next-generation Sequencing
- Thermo Fisher Scientific: Ion Torrent Next-Generation Sequencing Applications
- <u>Thermo Fisher Scientific: Introduction to Next-Generation Sequencing eBook</u>
- PacBio Sequencing Video How it Works
- <u>NIH | NLM Article PacBio Sequencing and Its Applications</u>
- Oxford Nanopore Technologies Video How nanopore sequencing works
- Oxford Nanopore Technologies Video Introduction to nanopore sequencing
- Oxford Nanopore Technologies Video Using nanopore sequencing: from first principles to applications
- Oxford Nanopore Technologies Video Update from Oxford Nanopore Technologies
- Bioinformatics Workbook GitHub Sequencing Technology
- Oxford Nanopore Technologies Video How to get started with nanopore sequencing and plan your experiment
- BMJ Article Four study design principles for genetic investigations using next generation sequencing
- ASM Training Videos Training in Infectious Disease Applications of NGS
- NGS Data Collection and Storage
- <u>ScienceDirect Article Next generation sequencing technology: Advances and applications</u>
- <u>NIH | NLM Article Next-Generation Sequencing and Emerging Technologies</u>
- ASM Training Videos Training in Infectious Disease Applications of NGS
 - Overview of Whole Genome Sequencing
 - Whole Genome Sequencing
 - Overview of Amplicon-Based NGS
 - Metagenomic NGS Methods
 - Overview of Shotgun Metagenomic Sequencing
 - <u>Next-Generation Sequencing in Microbiology (Part 1)</u>
 - Next-Generation Sequencing in Microbiology (Part 2)
- Chan Zuckerberg Biohub Rapid Response Resources
- <u>Slides Introduction to mNGS and lab setup</u>
- Slides Introduction to Illumina sequencing and data analysis for mNGS



References

- OpenAI. (2024). Gemini response on learning objectives for an introduction to genomic sequencing and NGS module. Retrieved July 29, 2024, from Gemini
- OpenAI. (2024). ChatGPT 4o mini response on learning objectives for an introduction to genomic sequencing and NGS module. Retrieved July 29, 2024, from ChatGPT
- OpenAI. (2024). Claude 3.5 Sonnet response on learning objectives for an introduction to genomic sequencing and NGS module. Retrieved July 29, 2024, from Claude
- OpenAI. (2024). Copilot response on learning objectives for an introduction to genomic sequencing and NGS module. Retrieved July 29, 2024, from Copilot



Acknowledgements

We would like to thank the following individuals, in alphabetical order of last name, for their valuable time and effort spent in designing (i.e., drafting, reviewing, and refining) this module: **Elizabeth Temiloluwa Akande, Temesgen Endalew, Siddiqah George, Carolina Matos, and Mohammed Ahmed Rameto**.

Furthermore, we would like to thank the following institutions, societies, journals, and individuals from whom we sourced open-access resources, used in this module:

American Society for Microbiology, Bioinformatics Workbook, British Medical Journal, Capacity Building for Bioinformatics in Latin America Project, Chan Zuckerberg Biohub, Chan Zuckerberg Initiative, Illumina, Instituto de Cálculo, National Institutes of Health | National Library of Medicine, Oxford Nanopore Technologies, Pacific Biosciences of California, ScienceDirect, Thermo Fisher Scientific; Kin Fai Au, Frederick Ausubel, Henk Buermans, Mark Cowley, Kevin Chau, Charles Chiu, Ryan Davis, John Dekker, Johan den Dunnen, Darío Fernández Do Porto, Xiaowu Gai, Andrew Gardner, Sissel Juul, Kishore Kumar, Paige Larkin, Clinton Mason, Nicholas Moore, Robin Patel, Melinda Poulter, Anthony Rhoads, Andrew Severin, Barton Slatko, Winston Timp, Dan Turner, Akelia Wauchope-Odumbo.