NGS 101 v2.0

A Re-introduction to the Fundamentals of Next-Generation Sequencing

AMP Association for Molecular Pathology

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COURSE DESCRIPTION:
NGS101 v2.0 course is an update of the very popular NGS101. Whether you're looking to add Next-Generation Sequencing (NGS) to your lab or need a refresher, this course delivers the most current and useful information to help you in your decision making process. This 7-part course (including 6.50 hrs of CME or CMLE credit) is a comprehensive primer on implementing and running NGS in your laboratory.

CERTIFICATE PROGRAM OUTLINE (The items in the red text are additional resources included in each chapter.)

Title	Speaker(s)	Description	Learning Objectives	Time to Complete
Welcome Remarks from Content Director	Joel A. Lefferts, PhD			4 min
Pre-Course Questionnaire				1 min
Pre-Test for Course				30 min
1: Value of NGS in Oncology and Germline Genetic Testing	Joel A. Lefferts, PhD	This presentation introduces next-generation sequencing and how it has revolutionized the field of molecular diagnostics. It covers the benefits as well as the limitations of the technology and, presents some case studies that showcases the use of NGS in both oncology and germline settings.	• Describe ways in which NGS has revolutionized molecular testing. • Explain the limitations and benefits of NGS vs other molecular testing options. • Define the main terminology used in various steps of NGS testing. • Identify ways that NGS results may be useful clinically for both germline genetic testing and somatic oncology testing.	60 min
Post-Test for Presentation 1				~5 min
2. Should I Consider NGS for my Laboratory?	Stephen J. Wicks, PhD	What should I consider to bring NGS in-house? Who should I involve and what is needed to build a business case? In this presentation learn about some of the important considerations to weigh when bringing NGS to your laboratory including IT infrastructure, bioinformatics pipeline, logistics, getting the right stakeholders involved, and more. Resource: a link to a paper and models to help you effectively communicate the cost and value of GSP services.	• Describe the use of NGS and its suitability for a laboratory. • List some of the steps involved in getting leadership buy-in and making a business plan for NGS. • Explain the infrastructure and requirements for an in-house NGS program. • Contrast the features of different models for deploying an NGS program. • Discuss potential issues with deploying an NGS program • Apply different resources to aid in deploying an NGS program.	33 min

Post-Test for Presentation 2				~5 min
3. Assay Design Considerations (How to Decide What is Best for my Lab?)	Matthew S. Lebo, PhD	This presentation will dive into the many questions to ask as you design your assay from the wet bench to the bioinformatics and provide tools and resources to help you in the design process. Resource: A worksheet with a list of links to suggested online tools and guidelines to help you get started on designing NGS assays.	• Define the components of an NGS assay, including the wet-laboratory portions and the bioinformatics processes. • Identify which NGS applications are suitable for specific testing scenarios. • Describe the considerations for interpreting and reporting findings from NGS assays.	52 min
Post-Test for Presentation 3				~5 min
4. How to Approach NGS Validation and Ensure Ongoing Quality	Susan J. Hsiao, MD, PhD	This presentation covers practical concepts in NGS assay validation from the wet lab to bioinformatics. Additionally, reviewed are elements of NGS quality assurance. Resource: Links to important clinical guidelines to help with validating your NGS assays.	• Describe the steps involved in validation of an NGS Assay. • Explain how to establish thresholds/cut-offs for quality metrics. • Identify when *in silico* vs wet validation is needed and also when a full validation or verification is sufficient.	56 min
Post-Test for Presentation 4				~5 min
5. I Found a Variant…Am I Finished? Variant Review (IGV, confirmation), Interpretation and Reporting	Matthew S. Lebo, PhD Honey V. Reddi, PhD	These two presentations will define the criteria for the evaluation and interpretation of germline/somatic NGS data and illustrate the components needed for reporting of clinical variants.	• Define the criteria for evaluation and interpretation of germline/somatic NGS data. • Illustrate the components needed for a germline/somatic report. • Describe the publicly available resources for germline/somatic NGS data analysis.	66 min
Post-Test for Presentation 5				~5 min
6. **What Might go Wrong?**	Joel A. Lefferts, PhD	In this presentation, learn about some pitfalls and challenges you may encounter in your NGS assay and how you might troubleshoot them. Resource: Link on standard nomenclature site.	• Recognize potential causes for false-positive or false-negative NGS results. • Identify sources for ensuring appropriate variant nomenclature. • Describe a situation in which manual review or re-validation of an NGS pipeline might be appropriate.	48 min
Post-Test for Presentation 6				~5 min
7. Advanced Topics, Future Directions a. Detection of Structural Variants by Targeted RNA Sequencing	Valentina Nardi, MD Nicole L. Hoppman, PhD Matthew S. Lebo, PhD Esther Babady, PhD,	These four presentations will cover advanced uses for NGS testing including RNA fusions, structural variants, infectious disease targets, and the bioinformatics involved in analyzing data from these uses.	• List alternative targets of NGS testing (e.g. RNA fusions, SV, ID targets). • Describe bioinformatics approaches needed for these alternative targets. • Explain some of the challenges of implementing NGS for these alternative targets.	75 min

b. Structural Variation – Why it's Important and How to Detect Using NGS c. Advances in Bioinformatics d. NGS in Infectious Diseases				
Closing Remarks	Joel A. Lefferts, PhD			1 min
Course Evaluation				5-10 min
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