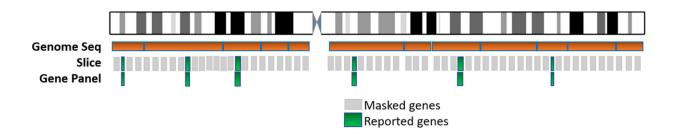


Best practice guidance for designing and utilizing slice testing approach for diagnostics

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Sequencing coverage of a representative chromosome for whole genome sequencing, exome sequencing, slice (green boxes indicating sliced regions analyzed) and gene panels. Credit: *The Journal of Molecular Diagnostics* (2023). DOI: 10.1016/j.jmoldx.2023.11.008

The Association for Molecular Pathology (AMP) today published a report that explores specific considerations for a slice testing strategy for diagnostics, including gene selection, analytic performance, coverage, quality, and interpretation.

The report offers expert consensus recommendations and results from an AMP-sponsored survey that compares a slice testing approach with traditional static gene panels and comprehensive exome analysis. The manuscript, "Slice Testing—Considerations from Ordering to Reporting: A Joint Report of the Association for Molecular Pathology, College of



American Pathologists, and National Society of Genetic Counselors," was released online ahead of publication in The *Journal of Molecular Diagnostics*.

As the number of <u>genes</u> associated with various germline disorders continues to grow, it is becoming more difficult for clinical laboratories to maintain separate assays for interrogating disease focused gene panels.

To help overcome this challenge, laboratories are employing a slice testing strategy that utilizes capture backbone to analyze data specific to a set of genes. Similar to traditional disease focused panels, a slice test sequences a broad set of genes but limits the interpretation to a predetermined list of relevant genes. This approach combines the advantages of high-quality gene panels with the flexibility and broad scope of exome sequencing.

"As with any emerging clinical methodology, information on slice testing is limited. This new report summarizes the current collective state of knowledge and assists clinical <u>laboratory</u> professionals with best practice guidance for test design and utilization," said Susan Hsiao, MD, Ph.D., 2023 AMP Clinical Practice Committee Chair and Associate Professor of Pathology and Cell Biology at Columbia University Vagelos College of Physicians and Surgeons.

"AMP will continue to work together with organizations like CAP and NSGC to monitor real-world evidence, share our members' expertise, and provide the broader laboratory community with a menu of guideline resources to help improve <u>clinical practice</u>."

The AMP Whole-Exome Sequencing Standards Working Group was established to survey current laboratory practices and develop initial best practice guidance for germline slice testing using exomes. The recommendations were informed by a directed review of existing



scientific literature, observational survey data, and the professional experience of the Working Group's subject matter experts.

The <u>report</u> outlines the capabilities, limitations, and considerations when using slice testing for laboratories and clinical providers. These expert consensus recommendations are meant to be a reference guide and not to be interpreted as a restrictive list.

"Slice testing is a relatively new practice that enhances flexibility in germline genetic testing for clinical laboratories and providers," said Rong Mao, MD, Chair of the AMP Whole-Exome Sequencing Standards Working Group and Medical Director of Molecular Genetics and Genomics at ARUP Laboratories. "AMP will continue to reassess and modify our best practice guidance as new data and/or reference materials become available."

More information: Jeffrey A. SoRelle et al, Slice Testing—Considerations from Ordering to Reporting: A Joint Report of the Association for Molecular Pathology, College of American Pathologists, and National Society of Genetic Counselors, *The Journal of Molecular Diagnostics* (2023). DOI: 10.1016/j.jmoldx.2023.11.008

Provided by Association for Molecular Pathology

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