

AMP Iissues consensus guideline recommendations for NGS bioinformatics pipelines

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The Association for Molecular Pathology (AMP), the premier global, non-profit molecular diagnostics professional society, today published 17 consensus recommendations to help clinical laboratory professionals achieve high-quality sequencing results and deliver better patient care. The report, "Standards and Guidelines for Validating Next Generation Sequencing Bioinformatics Pipelines: A Joint Recommendation of the Association for Molecular Pathology and College of American Pathologists," was released online ahead of publication in *The Journal of Molecular Diagnostics*.

"While the recent widespread adoption of next-generation sequencing (NGS) methodologies have transformed our ability to detect somatic and germline variants, the constant technology evolution and absence of professional guidelines have contributed to variability in clinical laboratory practice," said Somak Roy, MD, Assistant Professor of Pathology at University of Pittsburgh Medical Center, Working Group Chair and AMP Member. "To help solve this unmet need, AMP convened and led a multidisciplinary subject matter expert working group with representation from the College of American Pathologists (CAP), and the American Medical Informatics Association (AMIA) to summarize current knowledge, expose challenges and provide guidance on how to develop, implement and validate high-quality bioinformatics pipelines to ensure better overall patient care."



To achieve optimal NGS test quality, the new bioinformatics report emphasizes the critical role of the properly-trained molecular laboratory professional and recommends practical advice for laboratories regarding bioinformatics pipeline design, development and operation. This latest report completes the series of guidelines designed to improve the entire NGS workflow. It follows the previously published guidelines on interpreting oncology sequence variants and next-generation sequencingbased oncology panel validation. All three reports are based on evidence from a review of published literature, empirical data, current laboratory practice surveys, feedback from multiple public meetings, and expert professional experiences. Since these recommendations represent current best practice in a rapidly developing field, AMP anticipates a need for ongoing updates.

"AMP members are among the early adopters and users of NGS technology in a clinical setting and these best practice consensus standards and guidelines are based on our collective knowledge and expertise," said Alexis B. Carter, MD, Physician Informaticist at Children's Healthcare of Atlanta, Working Group Member, 2017 AMP Informatics Subdivision Chair, and AMP Board Member. "With this series of three guidelines addressing the complete NGS workflow, AMP has now provided the entire cancer genomics community with the appropriate tools and guidance to better incorporate the latest technological innovations in <u>molecular pathology</u>."

More information: *Journal of Molecular Diagnostics* (2017). <u>DOI:</u> <u>10.1016/j.jmoldx.2017.11.003</u>, jmd.amjpathol.org/article/S152... (17)30373-2/fulltext

Provided by Association for Molecular Pathology



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