

AMP reports on possibilities, challenges, and applications of next-generation sequencing

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Bethesda, MD, October 18, 2012 The Association for Molecular Pathology (AMP) published the report of the Whole Genome Analysis (WGA) Working Group of the AMP Clinical Practice Committee in the November 2012 issue of The *Journal of Molecular Diagnostics (JMD)*.

Titled "Opportunities and Challenges Associated with Clinical Diagnostic <u>Genome Sequencing</u>," the timely report provides a detailed and compelling overview of the landscape of next generation sequencing (NGS) technology and its clinical relevance and impact on improving patient care. The issues addressed in the report are comprehensive, including "...current technical, bioinformatic, and clinical implementation considerations, as well as medical applications, clinical utility, and ethical, legal, and education issues presented by genome-level diagnostic testing."¹

NGS-related technologies and instrumentation introduced over the past decade have significantly alleviated the amount of resources required to conduct complex genomic analysis. "The possibilities of these technologies are numerous," noted, Iris Schrijver, AMP President and lead author of the report. "As the only Association whose members are experts specifically dedicated to the field of <u>molecular pathology</u>, AMP has the responsibility to initiate and lead the discussion on safe, effective and practical applications of next-generation sequencing."

The report illuminates the current state of NGS by calling out and highlighting the issues to be addressed in greater depth moving forward.



"The next phase of reporting will include detailed studies of the validation, interpretation, and bioinformatics elements of NGS. We are excited to collaborate with other organizations to develop guidelines for application and quality assessment," said Jane Gibson, WGA Working Group Chair. "While this report is comprehensive, there is much yet to be done."

More information: A full copy of the report is available online at: <u>bit.ly/QVQSqt</u>

¹ Schrijver, I, et al: Opportunities and Clinical Challenges Associated with Clinical Diagnostic Genome Sequencing. *The Journal of Molecular Diagnostics* 2012, 14: 541-549.

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