JMD publishes article on laboratory perspective of incidental findings reporting

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The Association for Molecular Pathology (AMP), the premier global, non-profit organization serving molecular laboratory professionals, announced that *The Journal of Molecular Diagnostics* today published a Special Article titled, "Reporting Incidental Findings in Genomic Scale Clinical Sequencing - A Clinical Laboratory Perspective." This paper offers new and important perspectives from the laboratory highlighting the need for increased understanding and transparency of complex genomic testing. It also outlines important recommendations, including the need for laboratories to establish clear and patient-friendly policies for delivering ancillary information generated from genome-wide genetic tests.

A copy of the paper is available online at http://jmd.amjpathol.org/article/S1525-1578%2814%2900245-1/fulltext .

The AMP Incidental Findings Working Group, including authors of this paper, have closely followed the incidental findings debate since early 2013 when the American College of Medical Genetics (ACMG) published its guidelines on incidental findings reporting. These recommendations primarily focused on the content of secondary information obtained from genetic tests, and not the pitfalls of technology limitations, which has placed a significant burden on laboratories to educate patients as well as physicians about the strengths and limitations of genetic testing.

Lead author and Chair of the AMP Incidental Findings Working Group, Madhuri Hegde, Ph.D., from Emory Genetics Laboratory, Department of Human Genetics, Emory University, finds that patient and physician education about how genomic data are interpreted as well an appreciation for technology limitations placed on to the lab have been overlooked.

"Patients have a choice whether or not to receive additional information that may be available as a result of a genetic test that looks across an entire genome of DNA. In most cases, patients are interested in learning more, but it's critical that we educate them, as well as the ordering physician, about their options and what can and cannot be reported," noted Dr. Hegde. "While genetic testing technologies have revolutionized the way we diagnose and treat disease, we must appreciate the technical limitations that still exist today. If a report comes back with no known genetic abnormalities, it doesn't mean that a pathogenic variant might not exist - we just can't see it or interpret it from the data we have right now."

Laboratory regulation also plays an important role in how incidental findings are reported. Clinical Laboratory Improvements Amendments (CLIA) regulations, proficiency testing, lab accreditation, and other quality measures oversee and govern laboratories and the tests that they develop. If the U.S. Food and Drug Administration (FDA) steps in, as proposed in their recent draft framework, it could make access to important tests challenging.

"While we want to underscore the need for continued discussion among stakeholders to improve our understanding of the effect of different test result disclosure policies on patients, providers, and laboratories, we don't want to lose any progress that we have achieved since the completion of the human genome project," said Elaine Lyon, Ph.D., AMP Past President. "The proposed laboratory developed test regulation

framework imposes substantially new requirements on clinical laboratories, hospitals, physicians, and other health care providers. This interference with the practice of medicine poses significant impact on patient access to vital molecular testing services necessary for improving patient care."

Provided by Association for Molecular Pathology