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Experts collaborate on evidence-based somatic variant classification system

The Association for Molecular Pathology has published guideline recommendations that assess the status of next-generation-sequencing–based cancer tests and establish standardized classification, annotation, interpretation, and reporting conventions for somatic sequence variants.

The CAP provided liaison representation to the AMP Clinical Practice Committee Interpretation of Sequence Variants in Somatic Conditions Working Group, which developed the consensus recommendation. Michael Datto, MD, PhD, of Duke University School of Medicine, and Neal I. Lindeman, MD, of Brigham and Women's Hospital, were the CAP's representatives.

The guideline, "Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer: A Joint Consensus Recommendation of the Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists," was released online Dec. 16, 2016 ahead of publication in the January issue of the Journal of Molecular Diagnostics.

"These new recommendations resulted from the successful ACMG, AMP, and CAP efforts on germline variant interpretation and were additionally informed by the diverse perspectives expressed at the ASCO, AMP, and CAP Genomic Roundtable stakeholder discussions," Marina N. Nikiforova, MD, a member of the working group and chair of the 2016 AMP Clinical Practice Committee, said in a statement. She is a professor of pathology at the University of Pittsburgh Medical Center.

To help standardize the interpretation and reporting process, the AMP convened a panel of experts to develop a set of guidelines based on evidence from a comprehensive review of published literature, empirical data, current laboratory practice surveys, feedback from multiple public meetings, and their own professional experiences. The report proposed a four-tiered system to categorize somatic sequence variations based on their clinical significance in cancer diagnosis, prognosis, and/or therapeutics: variants with strong clinical significance (tier 1), variants with potential clinical significance (tier 2), variants of unknown clinical significance (tier 3), and variants deemed benign or likely benign (tier 4).

"We worked diligently to ensure the cancer genomics community was well represented," said Marilyn M. Li, MD, working group chair, "and it is our hope that we will soon see the widespread adoption of these guidelines leading to improved communication between molecular pathologists, oncologists, pathologists, and, most importantly, patients." Dr. Li is vice chief of the Division of Genomic Diagnostics and director of cancer genomic diagnostics at Children's Hospital of Philadelphia.

The article is at http://dx.doi.org/10.1016/j.jmoldx.2016.10.002. See next month's issue of CAP TODAY for a discussion of the consensus recommendation.

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FDA clears next-generation test for MRSA colonization

Cepheid received clearance from the Food and Drug Administration for its Xpert MRSA NxG, the next-generation methicillin-resistant Staphylococcus aureus infection control test. Xpert MRSA NxG is an on-demand molecular test that provides results in about one hour.

Xpert MRSA NxG was developed using an extensive library of MRSA strains collected from around the world and demonstrates unprecedented strain coverage, according to the company. Integral to the new test design are updated PCR primers and probes that detect both mecA and mecC strains, which reduces the frequency of false-positive results due to "empty cassette" strains. The test has been validated for use with ESwab (Copan) and rayon swabs.
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PMA approval granted for Aptima HIV-1 Quant

The Food and Drug Administration has granted PMA approval for the Hologic HIV-1 viral load monitoring assay. The Aptima HIV-1 Quant assay is a nucleic acid amplification test for the quantitative detection of RNA from HIV in plasma specimens. It runs on Hologic's Panther system.

The Aptima HIV-1 Quant assay is not approved for HIV-1 diagnosis in the United States. Outside the U.S., the Aptima HIV-1 Quant Dx assay is CE-IVD marked for diagnostic and monitoring claims.

The Aptima Quant viral load assays are not part of the pending sale of Hologic's blood donor screening business to Grifols, and the assays will continue to be owned by Hologic upon closing of the transaction with Grifols.
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Henry Ford announces automation partnership

Henry Ford Health System Pathology and Laboratory Medicine announced a partnership with Beckman Coulter that will fully automate the Henry Ford Hospital laboratory and modernize equipment at 12 other Henry Ford locations.

Henry Ford embedded automation into its Lean enterprise "refined over 12 years of training all laboratory employees to contribute in a culture of continuous improvement," says Richard Zarbo, MD, DMD, system chairman, Pathology and Laboratory Medicine Product Line. "Automation with a human touch" is what they call it, he says, adding that it "vastly improved workflow."

"For instance, in the nonautomated lab, 69 FTEs walked over 41 miles per day. The automated design saved 25.8 miles of walking," he says, "waste nearly equivalent to a marathon a day, or 1.5 FTEs doing nothing but walking."

The partnership with Beckman Coulter will enhance and streamline operations for greater efficiency.
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ArcherDX launches immune repertoire sequencing assays

ArcherDX has released the Archer Immunoverse immune repertoire sequencing assays. These targeted sequencing assays are the latest addition to the Archer product portfolio that now includes the Reveal ctDNA 28 assay for liquid biopsy research and the VariantPlex and FusionPlex assays for SNV/indel, copy number variation, and gene fusion detection.

ArcherDX announced Immunoverse at the American Society of Hematology annual meeting in December.

Immunoverse kits are targeted NGS assays to characterize the human immune repertoire from RNA. The assays use patented Anchored Multiplex PCR (AMP) chemistry for open-ended amplification from molecular-barcoded adapters. “We quickly found that AMP chemistry is ideal for interrogating VDJ recombination and gaining a true and reproducible measure of clonotypes,” Jason Myers, PhD, CEO of ArcherDX, said in a statement. “This unbiased approach towards VDJ-recombined sequences can yield more than 100,000 clonotypes in a single reaction, without the fear of PCR bias.”

The rearrangement of variable (V), diversity (D), and joining (J) gene segments, also known as V(D)J recombination, is the mechanism responsible for generating diversity in the antigen-recognition regions of B and T cell receptors.

Once sequenced, Immunoverse libraries are analyzed using the immune repertoire pipeline in Archer Analysis, a tool for clonotype identification and frequency reporting. Archer Analysis uses AMP-specific molecular barcode adapters to correct for PCR and sequencing errors.
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