ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

FOR IMMEDIATE RELEASE

Kathy Moran, MBA
Kmoran@acmg.net

Points to Consider in the Reevaluation and Reanalysis of Genomic Test Results: A Statement of the American College of Medical Genetics and Genomics (ACMG)

BETHESDA, MD – April 24, 2019—Advances in genetic testing have enabled clinical laboratories to provide increasingly comprehensive genetic testing. Now, in an effort to provide guidance on the important topic of the later reanalysis of genetic testing results, the American College of Medical Genetics and Genomics (ACMG) has published, "Points to Consider in the Reevaluation and Reanalysis of Genomic Test Results: A Statement of the American College of Medical Genetics and Genomics."

Why is the reanalysis of genomic testing results important? A steadily increasing amount of public and private variant information from affected and unaffected individuals, together with improved algorithms for predicting variant pathogenicity and an expanding body of functional studies, are creating a continuously evolving knowledge base for variant interpretation. Therefore, variant interpretation remains a dynamic process, and previously classified variants may benefit from periodic reanalysis. Subsequent reanalysis can also be critical in the search for diagnoses and for patient management as new gene-disease and variant-disease relationship are discovered. However, periodic reevaluation of the clinical significance of DNA variants also requires measurable time and financial resources from both laboratories and clinicians, for which there is currently no reimbursement paradigm.

The new ACMG Points to Consider document is specifically intended to "assist laboratories with the development of policies and protocols for variant reevaluation and case-level reanalysis appropriate for their setting and the specific circumstances presented by each clinical scenario."

ACMG President, Tony Gregg, MD, MBA, FACMG, FACOG said, "This document will guide patients, providers of care and laboratories at a time when genomic medicine is increasingly used in clinical practice. Guidance on reanalysis of genetic test results, especially those that involve next generation sequencing, is essential. Importantly, the ACMG Laboratory Quality Assurance Committee included specific points to consider for a variety of scenarios."

-more-

According to the ACMG Points to Consider, "Reanalysis practices should be designed to maximize clinical impact while minimizing the burden to the laboratory and healthcare system. (This) Points to Consider should be viewed as informative guidance for physician providers, clinical laboratory geneticists, genetic counselors and when appropriate, other health care providers."

The comprehensive "ACMG Points to Consider in the Reevaluation and Reanalysis of Genomic Test Results" includes:

-General Considerations
-Considerations for Variant-Level Reevaluation
-Considerations for Case-Level Reanalysis
-Considerations for Reporting
-Considerations for Retesting

It concludes, "Clinical laboratories should have policies and protocols for variant-level reevaluation and case-level reanalysis and these policies should keep pace with any new developments in population databases, genomic curation, bioinformatics and electronic health records. The efforts required to provide accurate and consistent variant classifications will be bolstered in the future by automation and changes in reimbursement. Ensuring that clinical laboratories can recoup the expense of this labor will be critical as the field evolves."

The new ACMG Points to Consider statement is available at https://www.nature.com/articles/s41436-019-0478-1.

-more-

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. The ACMG provides education, resources and a voice for more than 2,300 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. The College's mission is to develop and sustain medical genetics-related initiatives in clinical and laboratory practice, education and advocacy. Four overarching strategies guide ACMG's work: 1) to reinforce and expand ACMG's position as the leader and prominent authority in the field of medical genetics and genomics while educating the medical community on the significant role that genetics and genomics will play in understanding, preventing, treating and curing disease; 2) to secure and expand the professional workforce for medical genetics and genomics; 3) to advocate for the specialty; and 4) to provide best-in-class education to members and nonmembers. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (http://www.acmg.net/) offers resources including policy statements, practice guidelines, educational programs and a 'Find a Genetic Service' tool. The educational and public health programs of the ACMG are dependent upon charitable gifts from corporations, foundations and individuals through the ACMG Foundation for Genetic and Genomic Medicine.

-end-