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ACMG Updates Seminal Laboratory Standard on *CFTR* Variant Testing

Bethesda, MD –May 14, 2020 | It has been nearly two decades since the American College of Medical Genetics and Genomics (ACMG), together with the American College of Obstetricians and Gynecologists, made the landmark recommendation that cystic fibrosis (CF) become the first target of pan-ethnic universal carrier screening, with ACMG identifying a core panel of 23 pathogenic variants to be tested. Now, with the widespread availability and decreased cost of next-generation sequencing, it is possible to more easily interrogate all regions of the *CFTR* gene and correlate variants with phenotypes. This ability to move beyond targeted molecular testing methods for routine CF screening and diagnosis led to the ACMG Laboratory Quality Assurance Committee's release of a new key document: "[*CFTR* Variant Testing: A Technical Standard of the American College of Medical Genetics and Genomics \(ACMG\)](#)" published in ACMG's official journal, *Genetics in Medicine*.

This technical standard includes revised information about CF and the *CFTR* gene, new testing considerations and methodologies, and updated recommendations for the interpretation and reporting of test results. Written in clearly delineated sections, this important new resource will become a well-used reference by molecular genetics laboratories everywhere.

"Now that CF screening and diagnosis are established tests and next-generation sequencing is an established method in many clinical genetics laboratories, the ACMG wanted to update its technical laboratory standards for *CFTR* variant testing so that they would not only better reflect current laboratory practices but would also better enable future advancements in the field," said lead author Josh Deignan, PhD, FACMG. "Though it wasn't originally planned, it seems appropriate to have these updated ACMG standards published in May since May is also National Cystic Fibrosis Awareness Month."

The new ACMG document includes detailed sections on What to Test, How to Test and What to Report regarding *CFTR* variant testing.

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 professional organization solely dedicated to improving health through the practice of medical genetics and genomics, and the only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization. The ACMG is the largest membership organization specifically for medical geneticists, providing education, resources and a voice for more than 2,400 clinical and laboratory geneticists, genetic

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counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG's mission is to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. 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, including clinical research, while educating the medical community on the significant role that genetics and genomics will continue to 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 journal. ACMG's website (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.

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