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American College of Medical Genetics and Genomics Releases Policy Statement on Genomic Sequencing

BETHESDA, MD–March 29, 2012 | T**he American College of Medical Genetics and Genomics (ACMG)** has released an important new Policy Statement on Genomic Sequencing. Released at the ACMG Annual Meeting in Charlotte, NC on March 28, the Policy Statement titled *"Points to Consider in the Clinical Application of Genomic Sequencing"* outlines general principles that are recommended as genome-scale DNA sequencing begins to be applied in the care of patients. These new guidelines should help guide clinicians as they use such testing and address:

- The categories of patients and types of clinical situations for which such testing will likely be the most helpful
- What patients need to know about genomic sequencing before it is performed
- Principles that guide the use of information from genomic sequencing to improve patient care and medical decision-making

ACMG President Wayne Grody, MD, PhD, FACMG said upon the Statement's release, "The transition from traditional targeted gene testing to genome-wide analysis constitutes a genuine sea change in medicine, offering vastly enhanced diagnostic power along with unprecedented challenges in test interpretation and reporting. This policy represents the first professional guidance for appropriate integration of this new technology into clinical practice."

"Genomic sequencing is now transitioning from the research laboratory into the clinic and will have a transformative effect on healthcare," said Bruce R. Korf, MD, PHD, FACMG, one of the authors of the new Policy Statement and past-president of the ACMG. "This statement by the American College of Medical Genetics and Genomics will help set the context for clinical genomic sequencing to insure maximum value and quality or our patients and the public."

In defining the need for this document, ACMG said in the Policy Statement, "Major advances in DNA sequencing technology have made it possible to do large-scale sequencing, up to and including whole genome sequencing, in an effort to identify a gene mutation that may provide a diagnosis for a patient with an abnormal phenotype. This strategy offers potential advantages over classic approaches in which genes are analyzed individually, often over a long period of time and at substantial expense. As a result, there is considerable

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interest in offering genomic sequencing-based tests on a clinical basis. This document outlines points to consider in the clinical application of genomic sequencing to the detection of germ-line mutations. It is expected that this document will require revision as this rapidly changing field evolves."

One of the main conclusions/recommendations of the Policy Statement is that genomic sequencing data must be integrated in the context of the medical and family history of the individual patient to guide medical decision-making. This should be done by a health provider who is familiar with the enormously complex and voluminous data that is obtained from genomic sequencing.

The full policy statement is available on ACMG's homepage at www.acmg.net.

Note to editors: To arrange interviews with genetics experts on this and other related topics email Kathy Beal at kbeal@acmg.net.

About the American College of Medical Genetics and Genomics and The ACMG Foundation
Founded in 1991, the American College of Medical Genetics and Genomics (www.acmg.net) advances the practice of medical genetics by providing education, resources and a voice for more nearly 1600 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals committed to the practice of medical genetics and genomics. ACMG's activities include the development of laboratory and practice standards and guidelines, advocating for quality genetic services in healthcare and in public health, and promoting the development of methods to diagnose, treat and prevent genetic disease. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Find a Geneticist tool. The educational and public health programs of the American College of Medical Genetics are dependent upon charitable gifts from corporations, foundations, and individuals. The ACMG Foundation (www.acmgfoundation.org), a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics and genetic counseling in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics' mission to "translate genes into health" by raising funds to promote the profession of medical genetics and genomics to medical students, to support the development of practice guidelines for practicing physicians, to advance the awareness and understanding of medical genetics and genomics in the general public and much more.

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