

ACMG NEWS

For Immediate Release

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New ACMG Position Statement Addresses Clinical Utility of Genetic and Genomic Services: Genetic Information Provides Value to Patients, Families and Society

Bethesda, MD - March 16, 2015 – What is the value of finding out a definitive diagnosis for a genetic disease? In response to a recent Centers for Medicare and Medicaid Services (CMS) statement, a new Position Statement from the American College of Medical Genetics and Genomics (ACMG) provides extensive support for the Clinical Utility of Genetic and Genomic Services. In the broadest sense, “clinical utility” refers to the likelihood that a given intervention (in this case, genetic or genomic testing information) will lead to an improved health outcome. More specifically, a test can provide information about diagnosis, treatment, management, or prevention of a disease that will be helpful to a patient and their family.

ACMG believes there is great clinical value in arriving at a precise medical diagnosis, enabling, among other things, a disorder’s cause and prognosis, as well as frequently informing preventive and treatment options. ACMG considers there to be important clinical utilities related to genetic/genomic information in the three main areas of:

- Clinical Utility for Individual Patients,
- Clinical Utility for Families, and
- Clinical Utility for Society.

Dr. Gail Herman, FACMG, ACMG president said, "The value of a genetic diagnosis is immeasurable - not just psychologically but also financially. It often brings resolution to the costly diagnostic odyssey that is pursued - sometimes over the course of years - in a quest to establish a diagnosis. New DNA sequencing technologies are greatly expanding the number of causative genes known for genetic disorders. We are finding that for many rare genetic disorders identified in children, there is a new mutation not carried by either parent. This finding dramatically lowers the recurrence risk for future children and is extremely important for family planning. Further gene identification is a necessary first step for future development of specific treatment. Finally, for many adult disorders, such as inherited forms of cancer susceptibility, specific diagnosis enables testing other at risk family members and can often

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prevent disease or lead to earlier diagnosis with greatly improved prognosis."

ACMG Executive Director Michael S. Watson, PhD, FACMG stated, "Clinical Utility must take into account the profound value a diagnosis can bring to an individual, their family and society in general. Often a genetic diagnosis provides extremely valuable information not only to the patient but also to family members who are at risk for such disorders and who can take appropriate steps such as preventive measures or early treatment."

The new ACMG Position Statement, "Clinical Utility of Genetic and Genomic Services" states, "ACMG believes there is great clinical value in arriving at a precise medical diagnosis, enabling, among other things, a disorder's cause and prognosis, as well as frequently informing preventive and treatment modalities." It adds, "Advances in molecular diagnostic technology continue to expand the number of disorders that can be precisely diagnosed through such mechanism as sequencing or application of microarrays."

The new ACMG Position Statement is available at [https://www.acmg.net/docs/ACMG Position Statement Clinical Utility of Genetic and Genomic Services AOP.pdf](https://www.acmg.net/docs/ACMG_Position_Statement_Clinical_Utility_of_Genetic_and_Genomic_Services_AOP.pdf)

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About the ACMG and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (www.acmg.net) advances the practice of medical genetics and genomics by providing education, resources and a voice for more than 1750 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. ACMG is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. The College's mission includes the following goals: 1) to define and promote excellence in the practice of medical genetics and genomics and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics and genomics education to fellow professionals, other healthcare providers, and the public; 3) to improve access to medical genetics and genomics services and to promote their integration into all of medicine; and 4) to serve as advocates for providers of medical genetics and genomics services and their patients. *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 through the ACMG Foundation for Genetic and Genomic Medicine (www.acmgfoundation.org)

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