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American College of Medical Genetics and Genomics
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ACMG Releases New Position Statement on the Public Disclosure of Clinically Relevant Genome Variants

BETHESDA, Nov. 7, 2012 | The American College of Medical Genetics and Genomics (ACMG) released a new official **Position Statement on the Public Disclosure of Clinically Relevant Genome Variants**. This important new statement addresses the problems resulting from gene patent monopolies that have allowed some to develop proprietary databases of the clinical meaning of the variants in particular genes.

Michael S. Watson, PhD, FACMG, executive director of the ACMG explained, "The next phase of the human genome project, which is to annotate the human genome sequence with the clinical and biological meaning of the sequences and variants, will require capturing information from a very large number of people from diverse populations across the US and internationally. Information that informs us about the meaning of genome sequences should be in the public domain where it can be used for the benefit of all."

The just-released Position Statement says, "The American College of Medical Genetics and Genomics (ACMG) believes that gene testing and the clinical data on which genetic data are interpreted must remain widely accessible and affordable, and that the development and improvement of safe and effective genetic tests should not be hindered. Monopolistic practices that limit a given genetic test to a single laboratory are inconsistent with ACMG's goals of broadly accessible and affordable genetic tests."

Position Statement on the Public Disclosure of Clinically Relevant Genome Variants

Genetic and genomic tests are playing an increasing role in medical practice, enabling prevention, diagnosis, and management of both rare and common disorders and the analysis of genetic changes associated with cancer. Testing is done in commercial, academic, and hospital laboratories throughout the world. The clinical interpretation of rare variants requires access to data on clinical annotation, but some laboratories have maintained private databases that are not publicly available. In some cases, these are deliberately withheld from public access for business reasons; in other cases, laboratories that have focused on rare disorders have not taken the steps to make data publicly available. **The American College of Medical Genetics and Genomics (ACMG) believes that gene testing and the clinical data on which genetic data are interpreted must remain widely accessible and affordable, and that the development and improvement of safe and effective genetic tests should not be hindered. Monopolistic practices that limit a given genetic test to a single laboratory are inconsistent with ACMG's goals of broadly accessible and affordable genetic tests.**

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These practices:

- Limit the accessibility of competitively priced genetic testing services and hinder test-specific development of national programs for quality assurance.
- Limit the number of knowledgeable individuals who can assist physicians, laboratory geneticists and counselors in the diagnosis, management and care of at-risk patients.
- Place the laboratory that possesses the data in the position of practicing medicine, since only they have the ability to use data to inform medical decision-making.
- Inhibit the training of the next generation of medical and laboratory geneticists, physicians, and scientists.

Therefore, it is the ACMG's position that:

- Clinical data underlying genome annotation that informs the clinical interpretation of molecular variants are fundamental to the practice of genetic and genomic medicine.
- Withholding/restricting the use of such information impedes its integration into medicine.
- Interpretations of genomic variants should be informed by the best clinical information available to clinicians and scientists.
- Payers, regulators and providers should work to bring the clinical data into publicly available resources.

Approved by the ACMG Board of Directors on November 6, 2012

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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 1600 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals committed to the practice of medical genetics. 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 for Genetic and Genomic Medicine** (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 and Genomics' mission to "translate genes into health" by raising funds to promote the profession of medical genetics and genomics to medical students, to fund the training of future medical geneticists, to support best-practices and tools for practicing physicians and laboratory directors, to promote awareness and understanding of our work in the general public, and much more.

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