

ACMG NEWS

For Immediate Release

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ACMG Says ClinGen Will be Critical Resource for Interpretation of Genome-Scale Testing to Improve Patient Care

May 26, 2015 -- Bethesda, MD —Tremendous advances have been made in decoding the human genome in recent years but critical questions remain regarding what these variants mean and how they can be applied in clinical practice. In a comprehensive paper to be published in *The New England Journal of Medicine* on May 27, "ClinGen: The Clinical Genome Resource," a consortium including investigators from the American College of Medical Genetics and Genomics (ACMG) provide a detailed overview of ClinGen, an NIH-supported program to evaluate the clinical relevance of genetic variants for use in precision medicine and to increase the understanding of their role in human health and disease.

ACMG Executive Director Michael S. Watson, PhD, FACMG said, "With ClinGen and ClinVar now in place to manage the big data problem inherent in medical genomics, we are well positioned to enable extensive data sharing among laboratories and clinician. This is expected to dramatically improve our understanding of the clinical implications of genetic variation and its role in improving patient and population health."

ClinGen's mission is to build a genomic knowledgebase to improve patient care. Genetic clinicians and medical researchers hope to use information about genetic variants in a variety of ways including making better predictions about an individual's risk of disease, to developing tailored treatments and to improve clinical decision-making.

"We're dealing with massive amounts of information: more than 80 million genetic variants have been discovered to date, and for most of them, we have no clear understanding of their role in human health and disease," said Heidi Rehm, PhD, associate professor of Pathology at BWH and director of the Laboratory for Molecular Medicine at Partners HealthCare Personalized Medicine and lead author of the paper. As genetic sequencing becomes more common, interpreting data in a meaningful way and standardizing practices is imperative. The enormity of the situation is daunting, but the potential impact on patient care has immense implications."

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The paper in *NEJM* states, "...the American College of Medical Genetics and Genomics (a ClinGen grantee), working with members of the sequence and structural variant communities, have developed new standards for interpreting genetic variants. ClinGen is now working with laboratories to facilitate adoption of the new standards and openly share the basis of their pathogenicity assertions. This allows labs to resolve interpretation differences through expert consensus and applications of these standardized methods which will rapidly improve the use of genetic testing in the clinical setting."

One important part of ClinGen is [ClinVar](#), a publicly-accessible database launched in April 2013 at the National Center for Biotechnology Information (NCBI), which archives information submitted about genetic variants with medical relevance. Clinicians, researchers and patients can look up information on ClinVar to see what is currently known about a genetic variant. According to the paper's authors, "...the ClinVar database is ...being embraced by patients and physicians as a system for sharing up-to-date information on clinically reported genetic variants."

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

Founded in 1991, [ACMG](#) is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. The American College of Medical Genetics and Genomics (www.acmg.net) provides 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. The College's mission is to develop and sustain genetic initiatives in clinical and laboratory practice, education and advocacy. Three guiding pillars underpin ACMG's work: 1) Clinical and Laboratory Practice: Establish the paradigm of genomic medicine by issuing statements and evidence-based or expert clinical and laboratory practice guidelines and through descriptions of best practices for the delivery of genomic medicine. 2) Education: Provide education and tools for medical geneticists, other health professionals and the public and grow the genetics workforce. 3) Advocacy: Work with policymakers and payers to support the responsible application of genomics in medical practice. *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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