

## ACMG NEWS

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### ***Genetics in Medicine* Publishes Special Issue Dedicated to Genomics in Electronic Health Records: First Collection of its Kind**

September 19, 2013 –Bethesda, MD – Genetic tests can now tell us whether we are at increased risk of various cancers, heart or kidney disease, asthma and a number of other conditions. Other genetic tests can tell whether you will respond to certain medicines or be harmed by side effects linked to your genetic code. But harnessing that information to benefit individual patients and prevent illnesses in others will require that doctors have access to genomic information for each patient. As health records are converted to digital form, the most likely place to store and retrieve genomic information will be Electronic Health Records (EHR). But when and how that happens will depend on having good models to build upon.

Now, in the first collection of its kind, the October 2013 issue of *Genetics in Medicine*, the official peer-reviewed journal of the American College of Medical Genetics and Genomics, ([www.nature.com/gim/](http://www.nature.com/gim/)) provides a series of research articles detailing challenges and solutions for integrating genomic data into EHR. The issue features the insights of research teams actively engaged in integrating genomic medicine into clinical care. Most of the contributions derive from the experiences of individual sites that comprise the Electronic Medical Records and Genomics (eMERGE) Network, a national consortium funded by the National Institutes of Health, but additional perspective is provided by a commercial EHR vendor and by the Clinical Sequencing Exploratory Research (CSER) consortium, a cooperative group exploring applications of genomic sequencing.

“Our hope is that this issue of *Genetics in Medicine* will serve as a ‘how to’ and ‘what to think about’ for any group tasked with launching a genomics program and integrating this data into the EHR at the point of care,” said Joseph Kannry, MD, a board-certified internist and Lead Technical Informaticist of the Epic Clinical Transformation Group, Mount Sinai Medical Center, New York, NY. “This issue should serve as a reference point for many years to come.”

Dr. Kannry and co-editor Marc Williams, MD FACMG, director of the Genomic Medicine Institute, Geisinger Health System, Danville, Pa., steered the effort to organize contributions and together wrote the lead editorial. In it they state that, “Successfully integrating genomics into clinical

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care requires a vision, a strategy that will achieve the vision, and an actionable implementation plan.” The case studies provided in this special issue outline the following challenges and potential solutions:

**How can genomics be meaningfully incorporated into routine healthcare?** [Hartzler et al. doi: GIM.2013.127] describe how a broad range of parties, including institutional leadership, physicians, information technology staff, and patients must be included in the conversation if genomic medicine is to be successful. The article describes different ways to ensure support systems are in place when launching a genomic medicine project.

**How will genomic data be stored, processed, updated and retrieved?** [citation: Kho et al. doi: GIM.2013.131] examine data currently captured in EHR systems and compare that to genomic data. They look forward to the need for long-term storage and retrieval and how data can be accessed and compared across time and in changing clinical circumstances.

Likewise, [Chute et al. doi: gim.2013.121] discuss the opportunities for large data sets of genomic information to help detect new genomic risk factors and clinically important information not possible until recently. They identify gaps in standards for coding and transmission of data and propose solutions.

**How will genomic data first be used to help patients?**

We already know that genomic data can be used to predict response to drugs. Many of the most commonly prescribed drugs, such as the blood thinner warfarin and the statin drug simvastatin can have adverse side effects in people with certain genetic backgrounds. Vanderbilt University Medical Center (VUMC) has established a program known as PREDICT (Pharmacogenomic Resource for Enhanced Decisions in Care and Treatment). In this issue, [Peterson et al.] describe the successful integration of genomic information to help guide decisions about prescription drug choices.

**How will patients be involved in decisions about their genomic information?**

[Hazin et al doi: GIM.2013.117.] explore issues related to patients’ needs to have equitable access to genetic testing. The authors explore the need for accessible educational materials, as well as how to share information, balancing privacy and security, and suggest potential policy solutions.

**What does the future hold for the integration of genomic information into the EHR?**

Changing physician behavior and improving care requires Genomic Decision Support(GDS). [citation: Overby et al doi: GIM.2013.128] GDS uses genomic results to provide physicians with recommendations and suggested actions to take at the point of care. In their closing commentary [citation: ] Drs. Kannry and Williams note “that within the next few years, we will see researchers develop external CDS(Clinical Decision Support) capable of generating messages that trigger specific actionable items in a commercial EHR. Until standard representation of genomic results occurs, widespread adaptation of CDS by commercial EHRs will continue to be challenging regardless of value propositions by providers and patients.”

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“If we believe that there is information in the genome that is going to lead to more effective and safer therapies, we need to solve these issues,” said Williams. “We are basically trying to build a bridge over a canyon, and you can’t leave out any of the key structural elements and expect the bridge to hold together. We really need to solve these problems if we want to move to what some people are calling precision medicine.”

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**About the ACMG and ACMG Foundation**

Founded in 1991, the American College of Medical Genetics and Genomics ([www.acmg.net](http://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. 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](http://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