©American College of Medical Genetics and Genomics

IN THIS ISSUE

Building a conduit for clinical genomics

see cover

William Gropper, *Construction of the Dam* (study for mural, US Department of the Interior building, Washington, DC). 1965.18.11A-C. Smithsonian Art Museum, transfer from the US Department of the Interior, National Park Service.

In the 1930s, as the United States was struggling to recover from the Great Depression, a series of public-works projects were undertaken that provided infrastructure, which in turn led to the emergence of the United States as a world power. Hydroelectric projects, such as the ones depicted by artist William Gropper, sought to control devastating flooding and harnessed the power of rivers great and small to produc e inexpensive electrical power that was distributed through a network of public utilities, transforming the country.

Metaphorically, it is the task of researchers, clinicians, and informaticists to begin to harness the vast power of the genome. Our field must learn to take the data generated from sequencing the human genome and transform it into clinically useful information. Through the application of filters and protocols coupled to databases of annotated genes, we must strive to deliver clinically relevant information to patients and their providers via electronic health record systems. We envision the day when the flood of information is controlled and used to improve health outcomes for all through clinical utility and public health.

Building a dam also creates a reservoir that can be used for recreation, and, indeed, the early use of genomics has consisted of recreational dips into the gene pool.

We now know that dams bring not only benefits but harms, including disruption of ecosystems and, when breached, the very catastrophes they were meant to prevent. Likewise, we recognize the potential perils of genomics, including breaches of privacy, incidental findings, increased costs, and the real risk of inundating the health-care system with a flood of information it is ill prepared to deal with.

Gropper studied with Robert Henri and George Bellows, founding members of the "Ashcan School," which led to the first truly American artistic movement, Regionalism. His style was influenced by his work as a radical cartoonist, and—like that of other iconic Regionalists, such as John Stuart Curry and Thomas Hart Benton—his work always had an undertone of social commentary. *Construction of the Dam* is free of his usual depictions of injustice, instead showcasing the drive and intensity of the workers. Although those working to implement genomic medicine rarely exhibit the bulging muscles and hard hats of Gropper's construction gang, we approach our work with the same drive and intensity, infused with the Regionalists' sense of social justice for all. *—Marc Williams, MD, Guest Editor*

NEWS BRIEFS

Case study in clinical genetics offers lessons

The transition from sequencing the human genome for research purposes to offering whole-genome sequencing (WGS) for diagnostic purposes is now fully under way. But many questions remain about the value of such testing and what determines clinical utility. In a commentary in the 17 July 2013 issue of *Science Translational Medicine*, Jacobs and colleagues offer lessons learned from their experience setting up a genomics medicine clinic at the Medical College of Wisconsin and its Children's Hospital. The authors argue that WGS has a role in clinical medicine despite the lack of clear clinical utility in some cases. Three years ago, the group began

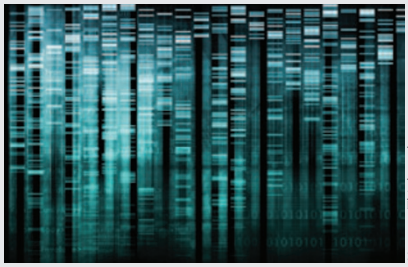
© istockphoto/Thinkstock

offering WGS to patients with undiagnosed rare diseases who had exhausted all standard care options. After 26 cases, the group reports a 27% success rate and a 39% failure rate. Ambiguous possible diagnoses make up the remaining 34% of cases. The authors say that more cases could be solved if sequenced human genomes were shared more widely among hospitals. They acknowledge the thorny issues of release of clinical data from hospital sites and informed consent. But in addition to those issues, the authors point to the lack of a standard data platform, which forces institutions to cobble together their own system of commercial and/or open-source software packages. They point out that current electronic health record systems can't handle complex medical data for patients with undiagnosed disease who have visited many clinicians and hospitals. Establishment of data and interoperability standards for genomic data therefore continues to be an important need. *—Karyn Hede, News Editor*

©American College of Medical Genetics and Genomics

NEWS BRIEFS *(continued)*

Consortium to push for global genomic data standards



Hemera/Thinkstock

We now take for granted that we can send a request for information on a computer server thousands of miles away and it pops up on our screens seamlessly. The now standardized and universal markup language that makes this possible was the product of the World Wide Web Consortium (W3C), the international organization that develops open standards for Web development. The ability to include genomic information in electronic health records will require similar standardized and accessible data. Now, a group modeling itself after W3C and representing 70 hospitals, research institutes, and technology companies from 40 countries has announced plans to develop the standards that would allow widespread genomic data sharing and the ability to combine genomic and clinical information. The group, called the Global Alliance, reports in a white paper available from the Massachusetts Institute of Technology's Broad Institute (http://www.broadinstitute.org/news/globalalliance) that they intend to create a common framework modeled after that of the W3C consortium. The group aims to promote open standards and best practices for organizations producing, using, or sharing clinical genomic data. By forging ahead of the coming onslaught of clinical genomic data, the group hopes to develop open standards that remove the incentives for creating competing, proprietary standards that have characterized early attempts to bring electronic health records to the United States. The organizations represented in the Global Alliance have pledged to protect individual privacy and to work toward harmonization of regulations governing access to data and results by participants, researchers, and other stakeholders. *—Karyn Hede, News Editor*

Genetics in Medicine | Mission Statement

Genetics in Medicine is a monthly journal committed to the timely publication of:

- Original reports which enhance the knowledge and practice of medical genetics
- Strategies and innovative approaches to the education of medical providers at all levels in the realm of genetics

As the official journal of the American College of Medical Genetics and Genomics (ACMG), the journal will:

- Provide a forum for discussion, debate and innovation concerning the changing and expanding role of medical genetics within the broader context of medicine
- Fulfill our responsibility to the College membership through the publication of guidelines, policy statements and other information that enhances the practice and understanding of medical genetics

Finally, as genetics becomes increasingly important in the wider medical arena, we will be an accessible and authoritative resource for the dissemination of medical genetic knowledge to providers outside of the genetics community through appropriate reviews, discussions, recommendations and guidelines.