

#### November 4, 2014

This month, I would like to bring to your attention a short new video that we recently completed, which 'tells the story' of NHGRI— our history, our organization, and our programs. This 'silent documentary' gives an overview of NHGRI through text, images, graphics, and video clips. I encourage you to visit <a href="youtube.com/watch?v=ktsNp6GeYGI">youtube.com/watch?v=ktsNp6GeYGI</a> to view the video.

In this issue of *The Genomics Landscape*, I describe the evolution of genome sequencing and its impact on public health surveillance and infectious disease diagnostics. I also highlight the first BD2K awards, National Family History Day, the appointment of a new NHGRI Executive Officer, as well as share details on how to comment on our recent genome sequencing workshop. See details below, along with other information items that I hope will be of interest to you.

Specifically, November's *The Genomics Landscape* features stories about:

- Changing the Face of Diagnostics in Clinical Microbiology
- First Big Data to Knowledge (BD2K) Awards
- Family Health History, the Original Genetic Test
- Appointment of Ellen Rolfes, NHGRI Executive Officer
- Comments Please: Report from the "Future Opportunities for Genome Sequencing and Beyond" Workshop

All the best,

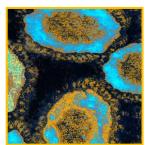


Watch here for current and upcoming locations of the Smithsonian-NHGRI exhibition "Genome: Unlocking Life's Code" as it tours North America!



# Changing the Face of Diagnostics in Clinical Microbiology

Healthcare-associated infections affect approximately two million people in the United States each year, resulting in ~100,000 deaths. Antibiotic-resistant infections cost the U.S. \$20-35 billion annually in healthcare expenses. Emerging infectious diseases need urgent containment and treatment. It is clear that in the U.S. and across the globe, we need better means to understand and control these infections.







From L to R: SARS-associated coronavirus, Klebsiella pneumoniae, and Ebola virus.

Recent public-health crises have brought to light the increasing real-time use of genomics in the area of infectious disease diagnostics. However, the potential role of genomics in this context was evident over a decade ago. For example, in 2003, Dr. Marco Marra and colleagues sequenced the genome of the SARS-associated coronavirus (Marra et al., Science 2003), aiding the understanding of the infectious outbreak in real time.

The advent of cheaper, faster DNA sequencing methods has created exciting new opportunities in the areas of public health and hospital epidemiology. Recently, NHGRI's own Dr. Julie Segre and colleagues used cutting-edge genomics to track an antibiotic-resistant strain of *Klebsiella pneumoniae* that was responsible for a deadly outbreak in the NIH Clinical Center (Snitkin et al., Sci Transl Med 2012). This past summer, Dr. Pardis Sabeti and colleagues sequenced the genomes of Ebola virus isolates from the recent outbreak in an effort to understand their origin and transmission (Gire et al., Science 2014).







Genome sequencing is serving many purposes in this rapidly-expanding area of infectious disease diagnostics. In addition to the above examples of patient-to-patient outbreak detection, new and unexpected microbes causing infection are being identified; investigators are finding genes conferring antibiotic-resistance or new

## First Big Data to Knowledge (BD2K) Awards



The new trans-NIH Big Data to Knowledge (BD2K) initiative, which aims to enable scientists to capitalize on biomedical 'Big Data' in advancing their research, recently announced the funding of its first set of grants. The BD2K Centers of Excellence, the LINCS-BD2K Perturbation Data Coordination and Integration Center, and the Data Discovery Index Coordination Consortium have started to receive funding - and exciting research is underway! Investigators from all of the funded BD2K programs will attend a joint kickoff meeting in November. For more information, see nih.gov/news/health/oct2014/od-09.htm.

## Family Health History, the Original Genetic Test



Health professionals have long known that many diseases run in families. This Thanksgiving, the U.S. Surgeon General encourages you to talk with your family about your family health history. The "My Family Health Portrait" online tool helps you organize your family health history and print it out to share with family members and your doctor(s). Updates to this tool are coming in November, including improved usability and optional additional information about your risk for colorectal cancer and type 2 diabetes. To learn more, visit <a href="https://hhs.gov/familyhistory">hhs.gov/familyhistory</a>.

modes of developing resistance; the origins of food-borne illnesses are being traced; reference genomes of clinical pathogens are being generated; the genomic variation of pathogens within patients is being investigated; and genome sequencing is being used for biodefense purposes, such as identifying specific genomic features of bacterial strains and tracing their origins via comparative genomics.

These developments have collectively pointed to the important role of genomics in infectious disease research and diagnostics. As a result, the President's Council of Advisors on Science and Technology (PCAST), an advisory group to the President consisting of top scientists and engineers in the U.S., recently prepared a <u>report</u> on combating antibiotic resistance. In the report, several actions were recommended: improving our surveillance capabilities, increasing the longevity of current antibiotics, and accelerating the rate at which new antibiotics and other interventions are discovered and developed. Genomics can contribute much to these efforts, especially surveillance and detection of resistance genes and strains, and is mentioned throughout the report.

# REPORT TO THE PRESIDENT ON COMBATING ANTIBIOTIC RESISTANCE



The PCAST report highlights the urgent need for additional research to develop approaches for combating antibiotic resistance. This issue surfaces in hospitals, homes, businesses, and schools across the world, and genomics has an important role to play in addressing these challenges. Large-scale genomic surveillance of pathogens, healthcare-related and otherwise, and the generation of reference microbial genome sequences will be crucial. The face of diagnostics and clinical microbiology is changing, and bright minds and improved technologies in genomics are helping to lead the way.







# Appointment of Ellen Rolfes, NHGRI Executive Officer



Last month, Ellen Rolfes, M.A., was appointed the NHGRI Executive Officer and Director of the Institute's Division of Management. This appointment followed a rigorous national search. In this position, Ms. Rolfes oversees all aspects of administrative management within NHGRI, including management analysis and evaluation, financial management, information technology, and human resources: she also serves as NHGRI's Deputy Ethics Counselor. Over her impressive career at NIH and NHGRI, Ms. Rolfes has worked in a variety of administrative management positions; most recently, she served as NHGRI's Deputy Executive Officer. To read more about Ms. Rolfes's appointment, visit genome.gov/27559294.

Comments Please: Report from the "Future Opportunities for Genome Sequencing and Beyond" Workshop



Last summer, NHGRI held a strategic planning workshop and program review to consider the future opportunities for genome sequencing, especially as it relates to the Institute's <u>Genome</u> <u>Sequencing Program</u>. A report from that workshop is now available, and we welcome your comments. To read the report and submit feedback, please see <u>genome.gov/27559219</u>.

#### **Genomics Research**

Advancing the Microbiome Research Community

<u>Nature Reviews Genetics 'Clinical</u>
<u>Applications of Next-Generation</u>
Sequencing'

Whole Exome Sequencing Closer to Becoming "New Family History"

Study: Most Respond Well to Genetic Testing Results

Exomes in the Clinic

<u>Thyroid Cancer Genome Analysis Finds</u>
Markers of Aggressive Tumors

<u>Autism Linked to Dozens of Genetic</u> <u>Mutations in Major New Studies</u>

Genome Advance of the Month

<u>Exploring Harmful Interactions Between</u>
<u>Artificial Sweeteners and Gut Microbiota</u>

**Genomics On Air** 

<u>Conversations on Health Care®: Eric</u> Green

#### **New Genomics Videos**

Genome: Unlocking Life's Code
Exhibition Closing Symposium —
Genomics and Global Health:
What Does the Future Hold

Genomic Medicine Centers VII
Meeting: Genomic Clinical
Decision Support

**NHGRI Showcase Video** 

#### **NIH News of Interest**

NIH Funds Research Consortia to Study More than 200 Rare Diseases

NIH Grantee Honored With 2014
Nobel Prize in Chemistry

NIH Awards ~\$31 Million to
Enhance Diversity in the
Biomedical Research Workforce

NIH Grants Examine How Genomic Information Can Affect Patients' Health

<u>Genomics Database Gets a</u> Patient Portal

### **Funding Opportunities**

Notice of Intent to Publish: Centers for the Genomics of Common Disease (UM1)

Notice of Intent to Publish: Centers for Mendelian Genomics (UM1)

NIH Director's Early Independence Awards (DP5)

NIH Offers Niche Assessment Program to SBIR and STTR Phase I Awardees

### **Funding News**

<u>Informational Webinar: 4D Nucleome</u> <u>Initiatives</u>

Webinars on NIH Application
Submission and Review (R15, Fs, SBIR/STTR & R01)

## **Media Availability**

TCGA Study Improves Understanding of Genetic Drivers of Thyroid Cancer

Researchers Uncover New Evidence
Revealing Molecular Paths to Autism

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