

# April 5, 2016

Spring has arrived, bringing a flurry of 'genomicists' to NIH for major talks. As part of the <u>NHGRI seminar series</u> celebrating the 25<sup>th</sup> Anniversary of the Human Genome Project's launch, Drs. Ewan Birney and Bob Cook-Deegan gave talks entitled "<u>Genomes and Big Data: A Personal View</u>" and "<u>Origins of the Human Genome Project: A Political History</u>," respectively. As the second installment of a special <u>Genomics and Health Disparities Lecture Series</u>, Dr. Neil Powe presented a talk entitled "<u>Chronic Kidney Disease: A Window into Understanding Health Disparities</u>." Finally, NIH hosted talks by Dr. Carlos Bustamante about "<u>Models and Data in Biomedicine: What's Real and What's Noise?</u> And Why Should We Care?" as part of <u>NIH's PI Day</u> events, and by Dr. Elaine Mardis about "<u>Translating the Cancer</u> <u>Genome: Transforming Cancer Care</u>" as part of the <u>NIH Director's Wednesday Afternoon Lecture Series</u>. Videoarchives of all five talks are available on NHGRI's <u>GenomeTV</u> or the <u>NIH Videocast website</u>.

For this issue of *The Genomics Landscape*, I am delighted to feature the cutting-edge behavioral research capabilities of the Immersive Virtual Testing Area (IVETA) found within our Institute's Intramural Research Program. This month's issue also highlights an educational infographic illustrating the basics of genetics and genomics, a study about DNA sequencing accuracy, and a new collaboration in genomic medicine.

Specifically, April's The Genomics Landscape features stories about:

- Virtual Reality Technology in Social and Behavioral Research
- Partnership for Community Outreach and Engagement in Genomics Infographic
- DNA Sequencing Accuracy: Sanger Versus Next-Generation Methods
- <u>NHGRI and Wellcome Trust Collaboration in Genomic Medicine</u>

All the best,

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



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To access past editions, see: <a href="mttps://genome.gov/27541196">genome.gov/27541196</a>

# Virtual-Reality Technology in Social and Behavioral Research

NHGRI's Intramural Research Program houses unique investigative endeavors of various types. One such enterprise is the Immersive Virtual Environment Test Unit, which encompasses the Immersive Virtual Testing Area (IVETA). Founded in 2006, IVETA is an experimental behavioral science research laboratory within the NIH Clinical Center that serves the NHGRI <u>Social and Behavioral</u> <u>Research Branch</u> (SBRB). The focus of IVETA is the development of advanced communications methodologies and technologies for use in behavioral research.



IVETA provides expertise, tools, and assistance to investigators who seek to integrate technologically-based behavioral research tools into their studies. The laboratory develops virtual-reality worlds for helping to answer research questions posed by SBRB investigators, and has recently expanded its scope to collaborate with other NIH researchers. IVETA's head is <u>Dr. Susan Persky</u>, who arrived at NHGRI in 2009. The laboratory uses innovative technologies, such as immersive virtual reality, for research projects that aim to advance knowledge at the intersections of genomics, human behavior, and society.



Dr. Susan Persky helps a volunteer using IVETA technology.

Dr. Persky and her collaborators are exploring how new genomic knowledge influences the interactions between healthcare providers and patients, as well as in other social circumstances. The IVETA technology uses virtual reality to simulate healthcare situations, examining how genomic information affects healthcare communication, relationships between healthcare providers and patients, medical decision-making, and peoples' emotions and behavior.

# Partnership for Community Outreach and Engagement in Genomics Infographic



NHGRI's Partnership for Community **Outreach and Engagement in Genomics** recently released a new infographic entitled Your Genome & You, which depicts some basic concepts about genetics and genomics for a public audience. The pictorialized lesson explains genetics, traits, and health, aiming to convey fundamental information about the importance of genetics and genomics in our everyday lives. For example, one of the sections of the infographic explains how the environment can influence how genes function. The Partnership for Community Outreach and Engagement in Genomics was established in 2014, and brings together community liaisons, health advocates, and health practitioners that together represent diverse populations. Their dedication to promoting a public understanding of genomics has been steadfast, and this infographic is a great addition to their previous and ongoing work. To access the infographic, see genome.gov/27564315.

# DNA Sequencing Accuracy: Sanger Versus Next-Generation Methods



As DNA sequencing gets increasingly used as a diagnostic tool in healthcare, it becomes more and more important to ensure that the results are correct. The classic and more expensive method of DNA sequencing – 'Sanger sequencing' – is now typically used to validate the more contemporary '<u>next-</u> generation sequencing' methods most often used for genome-sequencing studies. A recent paper by Dr. Les Biesecker in the NHGRI Intramural Research Program describes a study that compared the resulting DNA sequence data generated by the two methods. The study found that the two methods are very close with respect to More specifically, IVETA simulates clinical visits through the use of virtual-reality healthcare providers or patients. The simulations allow researchers to vary elements that they would like to study; for example, health disparities can be investigated by varying the race or ethnicity of the healthcare provider or patient, and the reactions of the test subject (who is in the other role) can be measured. In one <u>study</u> focusing on obesity, IVETA has been used as a behavioral measure for parents by allowing them to pick food for their children from a virtual-reality buffet and then measuring responses. This same approach could also be used to design a simulation for measuring a patient's own food choices.

In another example, a virtual-reality classroom was created to assess attention performance among children with Attention Deficit Hyperactivity Disorder. Such a simulation allowed researchers to administer realistic but controlled distractions (e.g., a bus driving by outside or a classmate whispering to another child at predetermined intervals) while child subjects were being studied. Researchers were also able to determine where in the classroom the child was looking throughout the assessment. Analyses of the data collected through this work are ongoing.



From left to right: virtual doctor, virtual buffet, and virtual patient.

This past year, the IVETA team undertook an initiative to widen the behavioral research tools being made available to investigators. They engaged in a horizon-scanning exercise and also held a series of meetings to discuss potential areas for expansion. In these meetings, they engaged both SBRB stakeholders as well as experts in behavioral science and information technology. Based on this effort, IVETA plans to expand its capacity to allow for other measurements that might complement wearable biosensors (such as those that capture participants' real-time cardiovascular activity). Additionally, the IVETA team is now evaluating the possible addition of automated analyses of facial expressions using webcam technology.

It is exciting to have something as novel as IVETA in NHGRI's Intramural Research Program. There are very few such 'immersive virtual reality' behavioral research laboratories in the world, and none that additionally focus on genomics. For more information about our IVETA program, see <u>genome.gov/27559802</u>. accuracy, with the 'next-generation' method actually coming out on top. These findings have potential implications for the current routine of relying on 'Sanger sequencing' methods to validate DNA sequencing results, especially in the context of clinical diagnostic testing. For more information, see genome.gov/27564480.

# NHGRI and Wellcome Trust Collaboration in Genomic Medicine



NHGRI's Clinical Genome Resource (ClinGen) is teaming up with the U.K. Wellcome Trust's Transforming Genetic Medicine Initiative (TGMI) to further research efforts for realizing genomic medicine. The two research groups met recently to coordinate their efforts in gathering evidence about the health relevance of genomic variants. Such evidence will be necessary for healthcare professionals to use genomic information in treating and managing patients. Meeting participants worked to define the approaches being used to evaluate health relevance of genomic variants, coordinate the assignment of disease areas being studied by each group to avoid duplication of efforts, share existing and planned platforms and tools, and define data standards. This new joint partnership will host a conference ("Curating the Clinical Genome") in June 2016 at the Wellcome Genome Campus in Cambridge, England. This meeting aims to convene the clinical genomics community to discuss best practices for the global sharing of clinical genomic data, with plans to focus on harnessing patient and population data for knowledge curation, using 'matchmaking' to discover new disease genes, and defining the evidence needed for establishing genedisease relationships.



Spotlight on the Precision Medicine Initiative (PMI)



nih.gov/precisionmedicine

- The <u>Funding Page</u> of the PMI Cohort Program website has been updated to list the awarded research projects.
  - From the White House Blog: "Your data in your hands: Enabling access to health information."

## **Genomics Research**

DNA Barcodes Could Streamline Search for New Drugs to Combat Cancer

Genes in Spotted Gar Could Boost Biomedical Use of Zebrafish

Advisory Committee to Address Framework for Building Medical Information Commons

Illuminating the Broad Spectrum of Disease

Single-Gene Mutations Account for Only 2 Percent of Cases of Severely Elevated Cholesterol

#### Genome Advance of the Month

Schizophrenia: Ancient Scrolls, Genomic Sequencing and Synaptic Pruning

#### **Request for Comment**

<u>NIH and FDA Request Public Comment</u> on Draft Clinical Trial Template for Phase 2 and 3 IND/IDE Studies

## **Resource & Funding Opportunities**

Gabriella Miller Kids First Pediatric Research Program Solicitation for Samples for Whole Genome Sequencing

NIH Director's Pioneer Award Program

# NIH/NHGRI News of Interest

The Big Data Quest to Treat Every Disease

Associate NIH Director for Biomedical Information Resources

Testimony on the FY2017 Budget Request Before the House Committee

Unlocking Life's Code: March 2016 Newsletter

National DNA Day: Improving Genomic Literacy on a National Scale

<u>NIH Leadership Co-Authors a Science</u> <u>Correspondence about the Importance of</u> <u>Basic Research</u>

Blue Ribbon Panel Announced to Help Guide Vice President Biden's National Cancer Moonshot Initiative

## **Funding News**

**Redesigned NIH Grants and Funding Site** 

Clarifications and Consolidated Biosketch Instructions and Format Pages Available for Applications with Due Dates on or After May 25, 2016

Reminder: NIH & AHRQ Grant Application Changes for Due Dates on or After May 25, 2016

## **Upcoming Twitter Chat**

National DNA Day Twitter Chat #DNADay16 – April 25

#### **Upcoming Webcasts**

<u>Genomic Medicine IX Meeting:</u> <u>Bedside to Bench – Mind the Gaps</u> – April 19-20

#### **New Videos**

<u>Genomes and Big Data: A Personal</u> <u>View</u> – Ewan Birney

Chronic Kidney Disease: A Window into Understanding Health Disparities – Neil Powe

Current Topics in Genome Analysis 2016

<u>Models and Data in Biomedicine:</u> <u>What's Real and What's Noise? And,</u> <u>Why Should We Care?</u> – Carlos Bustamante

Future Directions for Undiagnosed Diseases Research: UDN and Beyond

Origins of the Human Genome Project: A Political History – Bob Cook-Deegan

<u>Translating the Cancer Genome:</u> <u>Transforming Cancer Care</u> – Elaine Mardis

