



August 5, 2014

One of the most visible and rewarding areas of genomic medicine implementation involves the use of genomic approaches for solving medical mysteries. There are now numerous success stories in which researchers and doctors have used new genomic approaches to diagnose a patient's ultra-rare disease. In this issue of *The Genomics Landscape*, I describe a signature program that focuses on this area of genomic medicine— the NIH Undiagnosed Diseases Network. I also highlight a recent publication from the Human Heredity and Health (H3Africa) Initiative and a number of education and outreach activities, including a genomics symposium to foster relationships with Native communities, a celebration of the life of Henrietta Lacks, and a tour of the NHGRI-Smithsonian exhibition *Genome: Unlocking Life's Code* by none other than Dr. James Watson— Nobel Laureate, former NHGRI Director, and Human Genome Project champion.

Specifically, the August 2014 *The Genomics Landscape* includes:

- [Undiagnosed Diseases Network: Solving Medical Mysteries](#)
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All the best,

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## Undiagnosed Diseases Network: Solving Medical Mysteries

The NIH Intramural Research Program offers an environment that fosters innovation and high-risk, high-reward research, making it a home to myriad unique programs. An outstanding example of this is the Undiagnosed Diseases Program (UDP), launched in 2008 as a partnership among NHGRI, the NIH Clinical Center, and the Office of Rare Diseases Research (now part of the National Center for Advancing Translational Sciences). The UDP's goals are to provide answers to patients with mysterious conditions that have long eluded diagnosis and to advance medical knowledge about rare and common diseases. To date, the UDP has accepted ~750 patients. In addition to robust clinical evaluations, many of these patients received cutting-edge genomic analyses, such as scans for single-nucleotide polymorphisms and whole-exome sequencing. The multidisciplinary clinical and research team has diagnosed ~100 patients, discovered 2 previously unknown diseases, and identified 50 genes not previously associated with any other human disease.



The success of the UDP was recognized by the collective NIH leadership in 2012, when it decided that the NIH Common Fund would provide over \$120 million over five years to build upon the success of the NIH UDP to create a network of medical research centers around the country, the Undiagnosed Diseases Network (UDN). The UDN's focus will be to continue studying patients with undiagnosed conditions, while promoting the use of genomic approaches for clinical diagnostics and engaging basic researchers to identify the underlying mechanisms of disease. Training clinicians in the use of contemporary genomic approaches is also an important component of the UDN. NHGRI Extramural Research Program staff members are playing a key role in leading this new NIH Common Fund program.

Since its launch, the UDN has released five Funding Opportunity Announcements (FOAs). The Coordinating Center for the UDN was awarded to Harvard Medical School (Principal Investigator Isaac Kohane, M.D., Ph.D.). Funds were also provided to support a number of gene function studies that are investigating rare and

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### *H3Africa Initiative Publishes Marker Paper*



The Human Heredity and Health in Africa (H3Africa) Initiative, an NIH Common Fund project co-led by NHGRI, recently published a 'marker' paper describing its plans for facilitating the development of genomics research in Africa. The project's efforts will support the development of contemporary research approaches to the study of genomics and environmental determinants of common diseases, with the goal of improving the health of African populations. To learn more about H3Africa, see [h3africa.org](http://h3africa.org). To access the paper, "Research Capacity: Enabling the Genomic Revolution in Africa," see [ncbi.nlm.nih.gov/pubmed/24948725](http://ncbi.nlm.nih.gov/pubmed/24948725).

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### *Brooklyn Community Celebrates Henrietta Lacks*

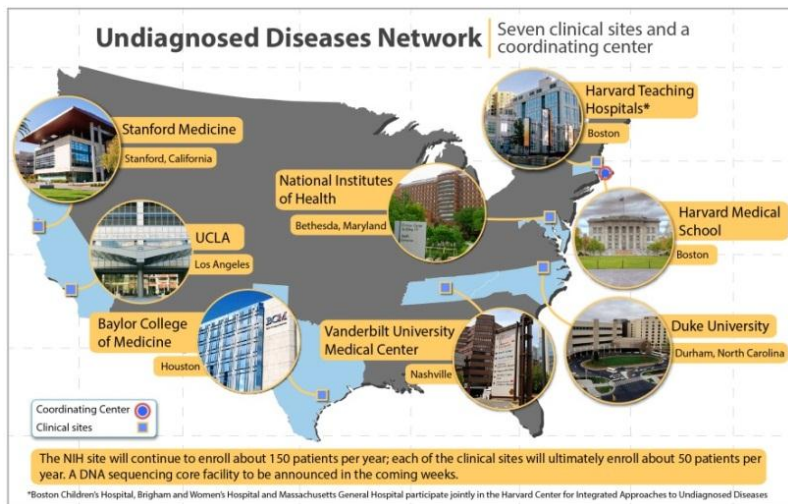


In mid-May, NHGRI partnered with the Brooklyn Public Library and the Smithsonian's National Museum of African American History and Culture to honor the legacy of Henrietta Lacks at a community-based event, "We Speak Your Name: Exploring the Life of Henrietta Lacks 'Heroine of Modern Science & Medicine.'" Students, educators, and audience members representing diverse cultures and socioeconomic demographics met at the library to examine important questions in ancestry, genomics, and culture, and to explore issues of race, class, gender, and bioethics through the story of Mrs. Lacks. Members of the Lacks family were present as the community honored Mrs. Lacks and publicly recognized her life and tremendous contributions to science and medicine. For more information, see [genome.gov/27558032](http://genome.gov/27558032).

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new diseases. Most recently, six awards were made to medical centers across the country (see map below) that will serve with the NIH UDP as the seven clinical sites for the UDN (see [genome.gov/27557990](http://genome.gov/27557990) for details). The Coordinating Center has already begun laying the groundwork for the seven UDN clinical sites to begin accepting patients.

New components will soon join the existing parts of the network. Specifically, facilities for genome sequencing and pilot projects for pursuing gene function studies will be funded before the end of this fiscal year, and another core laboratory funding announcement is forthcoming. We are well on our way to seeing this complex network of resources and expertise come to fruition.



Map showing the newly awarded UDN clinical sites and Coordinating Center.

The NIH UDP— and now the UDN— are exceptional examples of genomic medicine in action. Right here on the NIH campus, and now at six other medical centers across the U.S., we have the ability to help children and adults (and their families) who have endured long diagnostic odysseys, many of whom had lost hope for understanding their disease before learning about these unique programs. Not only will the UDN provide tangible medical benefits, but its centers will develop common protocols, operating guidelines, and practices that can be shared broadly, along with systems for data collection and common approaches for patient selection, evaluation, and diagnosis. The era of genomic medicine has arrived!

For more information on the UDN, see [commonfund.nih.gov/Diseases/index](http://commonfund.nih.gov/Diseases/index).



### Symposium Held to Explore Native Communities' Perspectives on Genomics



In late June, NHGRI joined the National Congress of American Indians and the Smithsonian's National Museum of the American Indian (NMAI) to co-host a symposium entitled "A Spectrum of Perspectives: Native Peoples and Genetic Research" at NMAI in Washington, D.C. At the symposium, there were thoughtful discussions about Native health, genomics and ancestry, training for Native researchers, and the ethics of Native blood collection and usage. Speakers represented numerous tribes from across the continent. For more information, see [genome.gov/27558499](http://genome.gov/27558499).

### Genome: Unlocking Life's Code Exhibition's Notable Visitor



Recently, Dr. James Watson, Nobel Laureate and former NHGRI Director, toured *Genome: Unlocking Life's Code*, a genomics exhibition developed through a partnership between NHGRI and the Smithsonian Institution's National Museum of Natural History (NMNH) in Washington, D.C. Dr. Watson and his co-discovery of DNA's double-helical structure are featured in the exhibition. The exhibition opened in June 2013, and its last day at the NMNH will be September 1<sup>st</sup>. For more information about the exhibition, see [unlockinglifescode.org](http://unlockinglifescode.org).



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

[Take Part in the Social Genomics Project](#)

[NIH Funds Next Step of Cutting-Edge Research into Alzheimer's Disease Genome](#)

[Unveiling a New Look – and More – for PubMed Commons](#)

[Autism Architecture: Unrolling the Genetic Blueprint](#)

[FDA Takes Steps to Help Ensure the Reliability of Certain Diagnostic Tests](#)

[Statement on FDA's Proposed Oversight of Laboratory-Developed Tests](#)

[Genotype-Tissue Expression Project Expands Functional Studies of Genomic Variation](#)

[NIH Awards Two New Grants to Explore the Understanding of Genomics Research in Africa](#)

[NIH Awards \\$14.5 Million to Research Groups Studying Newest DNA Sequencing Techniques](#)

### ***NIH Funding News***

[Formula for Innovation: People + Ideas + Time](#)

### ***Genomics Research News***

[Gut Bacteria Linked to Rheumatoid Arthritis](#)

['Unloved' Pseudogenes May Provide Clearer Understanding of Biomarkers](#)

[Francis Collins Says Medicine in the Future Will Be Tailored to Your Genes](#)

[Study Identifies Novel Genomics Changes in the Most Common Type of Lung Cancer](#)

[Bioinformatics: Big Data Versus the Big C](#)

[Scientists Identify Gene Linked to Fatal Inflammatory Disease in Children](#)

[Exhibit on Native Health Comes to Honolulu](#)

[Common Gene Variants Account for Most Genetic Risk for Autism](#)

[Schizophrenia's Genetic Skyline Rising](#)

[The Cancer Genome Atlas Researchers Identify Four Subtypes of Stomach Cancer](#)

[Scientists Find Six New Genetic Risk Factors for Parkinson's](#)

[Smoking Mothers May Alter the DNA of Their Children](#)

[Marmoset Sequence Sheds Light on Primate Biology, Evolution](#)

### ***Genome Advance of the Month***

[Improving the Detection of Heart Transplant Rejection with DNA Sequencing](#)

### ***New Genomics Videos***

[A Spectrum of Perspectives: Native Peoples & Genetic Research](#) – June 27

[Future Opportunities for Genome Sequencing and Beyond: NHGRI Planning Workshop](#) – July 28-29

### ***Workshops***

[Assessing Genomic Sequencing Information for Health Care Decision Making – IOM Workshop Summary](#)

### ***Funding Opportunities***

[Ethical, Legal, and Social Implications \(ELSI\) of Genomic Research Program](#)

[NIH Transformative Research Awards](#)

[The Electronic Medical Records and Genomics \(eMERGE\) Network, Phase III RFAs](#)

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