May 5, 2015

The recently announced Precision Medicine Initiative is on the minds of many people these days. Numerous developments are unfolding as the planning phase proceeds, and it seems that everywhere that I go, scientists and non-scientists alike are eager to hear details. Accordingly, I have added a new 'spotlight' on the Precision Medicine Initiative within *The Genomics Landscape*—immediately before the section containing links of interest. Each month, this spotlight will feature news regarding the Precision Medicine Initiative, so stay on the lookout for these regular updates.

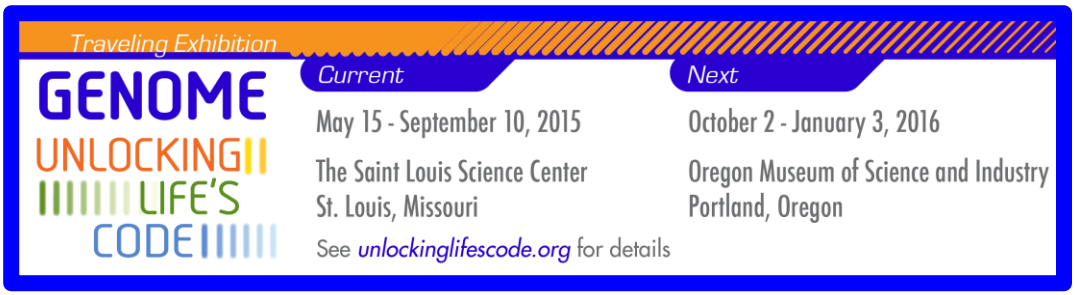
In this month's *The Genomics Landscape*, I discuss the NHGRI-EBI Genome-Wide Association Studies Catalog, which was started at NHGRI and is now housed at the European Bioinformatics Institute (EBI) in the U.K. See various details below, along with other information items that I hope will be of interest to you.

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

- Genome-Wide Association Studies Catalog
- G2C2: Updated Genetic Counselor Resources Now Available
- NHGRI Resources Regarding Investigational Device Exemption (IDE) Regulations
- Genome: Unlocking Life's Code–Spring Events
- NHGRI Staff Visit NASA

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!



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~To suggest future topics, send an e-mail to: NHGRILANDSCAPE@MAIL.NIH.GOV~
~To access past editions, see: genome.gov/27541196~

Genome-Wide Association Studies Catalog

Genome-wide association studies (GWAS) have proven to be effective for identifying and understanding the genetics underlying human health and disease. The GWAS approach takes advantage of high-throughput DNA analysis methods to examine common genomic variants across human genomes; the goal is to find the variants associated with particular diseases or traits. Once detected, scientists can further study the variants (or variants nearby) to determine their role, if any, in causing disease or influencing the relevant trait.

NHGRI launched the Genome-Wide Association Studies (GWAS) Catalog in 2008 to capture information about published GWAS efforts and to create a curated, downloadable, and user-friendly data resource. The GWAS Catalog started as a series of tables that evolved into a database and the now-iconic chromosome-based graphic seen below. In 2010, NHGRI began a fruitful collaboration with the European Molecular Biology Laboratory-European Bioinformatics Institute (EMBL-EBI), in the U.K., to improve the Catalog experience through additional curation, automated updating, and enhanced search capabilities. Very recently, the GWAS Catalog moved to the EMBL-EBI, but will continue to be curated jointly by EMBL-EBI and NHGRI staff.

GWAS Catalog graphic from May 5, 2015 (ebi.ac.uk/fgpt/gwas/images/timeseries/gwas-latest.png).

To manage and maintain a high-quality data set, Catalog curators developed specific inclusion criteria. Studies must assay a minimum number of single-nucleotide polymorphisms (SNPs), a type of genomic variant, and include SNP-trait associations that achieve a certain level of statistical significance. The database is highly functional, with an interactive graphic and facile search functions. Users can search the Catalog based on different attributes (e.g.,

G2C2: Updated Genetic Counselor Resources Now Available

G2C2 GENETICS/GENOMICS COMPETENCY CENTER

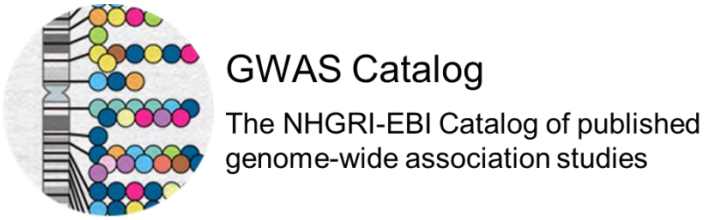
The Genetics/Genomics Competency Center for Education (G2C2)—a free, online collection of materials for self-directed learning in genetics and genomics—provides high-quality educational resources for genetic counselors, nurses, physician assistants, pharmacists, and physicians. In April, the Genetic Counselor resources on G2C2 were updated based upon the revised practice-based competencies announced by the Accreditation Council for Genetic Counseling (ACGC). The updated genetic counselor portal on G2C2 focuses on web-based content and interactive learning programs. Groups are encouraged to submit new genetic counseling resources to G2C2; it's a great way to reach the genetic counseling community and beyond.

NHGRI Resources Regarding Investigational Device Exemption (IDE) Regulations

In order to help the research community understand and navigate the Food and Drug Administration's (FDA) Investigational Device Exemption (IDE) regulations, NHGRI has developed a number of resources available on genome.gov. These include a plain-language guide for researchers that explains when the regulations might apply to their genomic medicine studies and a series of case studies. Further case studies will be developed as more projects work their way through the FDA system.

author, gene, and SNP) and traits (e.g., cancer, diabetes, and obesity).

The real power of the GWAS Catalog lies in its utility. It is gratifying to see researchers incorporate the GWAS Catalog into other genomic resources and use the data to further research studies. GWAS Catalog data have been incorporated into a number of genome browsers, including dbSNP, UCSC, and Ensembl. It has also spawned the development of the Phenotype-Genotype Integrator (PheGenI), a resource developed with NCBI to link GWAS Catalog data with other NCBI databases. NHGRI's Encyclopedia of DNA Elements (ENCODE) Project utilized the GWAS Catalog to identify regulatory elements associated with human disease.



The GWAS Catalog continues to be a valuable resource for information regarding the vast number of findings coming out of GWAS. With the last data release on May 2, 2015, the GWAS Catalog contained 2,154 published studies and reported information about 15,333 SNPs. The GWAS Catalog graphic has become an iconic image that I see often used to represent the progress made in common disease-associated variant discovery. NHGRI and EBI are committed to maintaining this resource as it continues to grow and serve the research community. To access the GWAS Catalog, see ebi.ac.uk/gwas/. Additional information is available at the NHGRI's GWAS web page: genome.gov/gwastudies/.

Genome: Unlocking Life's Code–Spring Events

GENOME UNLOCKING LIFE'S CODE

The *Genome: Unlocking Life's Code* exhibition ended its tenure at The Tech Museum of Innovation in San Jose, CA on April 27. The exhibition is now en route to the Saint Louis Science Center in St. Louis, MO, where it will open on May 15. During the exhibition's stay in San Jose, several events were held to promote participation from San Francisco Bay Area students, residents, researchers, and healthcare providers. In March, Drs. Jean Jenkins and Kathleen Calzone hosted free educational programs for practicing nurses and nursing educators to highlight the value of the exhibition as an innovative teaching tool. In early April, Dr. Eric Green moderated discussions with Bay Area research leaders from both the private and academic sector who are at the forefront of research on big data, genomics, and precision medicine as part of the "Big Data, Genomics, and Precision Medicine" symposium. Most recently, on April 18 the museum hosted "Epic Genetics Day", which included hands-on activities to explore how DNA works and a presentation by NHGRI's Dr. Elaine Ostrander. For further details about the traveling exhibition and future events, see unlockinglifescode.org.

NHGRI Staff Visit NASA

Earlier this year, NHGRI staff members Drs. Lawrence Brody, Sara Hull, and Jean McEwen were invited to the Johnson Space Center in Houston, TX by NASA's Human Subjects Research team. Those assembled discussed issues related to genomics and NASA human biology research programs, including policies related to informed consent, vulnerable subjects, privacy, and data sharing. NASA aims to understand the challenges that astronauts face in space through programs such as the Twins Study, which will employ genome sequencing.

Shown left to right: Drs. Lawrence Brody, Lee Morin (space shuttle astronaut), and Jean McEwen at the Johnson Space Center.

Group photo in historic Apollo-era Mission Control center at the Johnson Space Center.

Dr. Sara Hull at Mission Control.

Spotlight on the Precision Medicine Initiative

- A group of experts has been assembled to construct a vision for the Precision Medicine Initiative (PMI), incorporating input from the public. Constituted as a Working Group of the Advisory Committee to the NIH Director, the group will also delineate important scientific questions that can be investigated by using a cohort of this size. The membership of this new Working Group is posted at nih.gov/precisionmedicine/working-group.htm.
- NIH released a Request for Information about the NIH Precision Medicine Cohort, with responses due by May 7. More information can be found at grants.nih.gov/grants/rfi/rfi.cfm?ID=43.
- A series of workshops are planned to collect input from participant, scientific, and other stakeholder groups as the PMI planning phase intensifies. For details on upcoming events and videocast information on past and future events, see nih.gov/precisionmedicine/events.htm.

Genomics Research

NIH Study Finds Genetic Link for Rare Intestinal Cancer

How Modern Life Depletes Our Gut Microbes

New Method Increases Accuracy of Ovarian Cancer Prognosis and Diagnosis

Zebrafishing for a Weapon against Metastatic Cancer

Genome Advance of the Month

Iceland Study Provides Insights into Disease, Paves Way for Large-Scale Genomic Studies

Upcoming Webcasts/Webinars

The Cancer Genome Atlas' 4th Scientific Symposium – May 11 & 12, 2015

Seventy-Fourth Meeting: National Advisory Council for Human Genome Research – May 18, 2015

GTEx Symposium: All Things Considered – Biospecimens, 'Omics Data, and Ethical Issues – May 20 & 21, 2015

NIH/NHGRI News of Interest

Whitescarver Steps Down as Director of NIH's Office of AIDS Research

New NIH Web Portal on Rigor and Reproducibility Launched

Photojournalist Bartlett Ends 35-Year NIH Career

Top Boards in the 2015 Unlocking Life's Code Pinterest Challenge

NIH Names Dr. Eliseo Pérez-Stable Director of the National Institute on Minority Health and Health Disparities

Using Genetics to Fight Disease – Interview about the Precision Medicine Initiative with Dr. Eric Green

Statement on NIH Funding of Research Using Gene-Editing Technologies in Human Embryos

Appointment of Adrienne A. Hallett as the Associate Director for Legislative Policy and Analysis, NIH

Service to America Medal Finalists – The Cancer Genome Atlas

Requests for Information

Optimizing Funding Policies and Other Strategies to Improve the Impact and Sustainability of Biomedical Research

Funding Opportunities

Genome Sequencing Program Coordinating Center

Centers of Excellence in Ethical, Legal, and Social Implications Research

Undiagnosed Diseases Gene Function Research

Funding News

Use of Cloud Computing Services of Controlled-Access Data Subject to the NIH Genomic Data Sharing (GDS) Policy

Potential Delays to NIH Issuing Awards in May 2015

Publication of the Revised NIH Grants Policy Statement

Reporting Publications in the Research Performance Progress Report

NIH Policy on Application Compliance

Racial and Ethnic Categories and Definitions for NIH Diversity Programs and for Other Reporting Purposes