April 1: Dr. Ada Hamosh and Dr. Nara Sobreira, PhenoDB: A Tool for Collection and Analysis of Phenotypic and Genomic Data

SYNOPSIS:

PhenoDB is a web-based tool developed for the collection, storage and analysis of phenotype data, as well as interpretation of exome and genome data in the context of phenotype data. It has it own taxonomy and links to OMIM for disease terms. There is a single center version that allow identifiers and includes only the phenotype and analysis module (http://phenodb.org) and a tool for larger studies that also includes a sample module and an ELSI module for storage and review of consents (http://researchphenodb.net). Both are freely available for download; http://phenodb.org can be used by individual users to try it out (it is toggled to have only deidentified data). PhenoDB has been in use for the Baylor-Hopkins Center for Mendelian Genomics since March 2012 and holds information on over 5000 individuals from ~3000 families. It has proved efficient and effective in novel disease gene discovery. It can also be used for a laboratory or clinic.

Session details...

BIOs:

Dr. Ada Hamosh, Dr. Frank V. Sutland Professor of Pediatric Genetics, is the Clinical Director of the McKusick-Nathans Institute of Genetic Medicine at the Johns Hopkins University School of Medicine, the Scientific Director of Online Mendelian Genetics in Man (OMIM®), and the Co-Chair of the Phenotype Review Committee of the combined Baylor-Hopkins Centers for Mendelian Genomics (CMG), a National Human Genome Research Institute-funded project to identify the genes responsible for known and novel Mendelian disorders. Over the past year, Dr. Hamosh and colleagues have developed PhenoDB, a Web-based tool for the collection, storage, and analysis of standardized phenotype and genotype data for use in the CMG project that is freely available to all for clinical and research use.

Dr. Hamosh has authored over 70 papers and serves on several international committees representing genome-phenome relationships, as well as phenotype ontologies, including the International Rare Disease Research Consortium, the Human Variome Project, the ClinGen Project, Global Alliance for Genomic Health, and the Human Genome Organization.

Dr. Nara Sobreira is an M.D. clinical geneticist, board certified in Brazil and board eligible in the U.S. She completed her Ph.D. in Human Genetics at Johns Hopkins in the laboratory of Dr. David Valle. While working on her Ph.D. and completing a medical genetics residency at Johns Hopkins, she has worked a science writer with OMIM since 2009 and will become the Deputy Scientific Director for Phenotypes for OMIM in July 2015, when she joins the faculty of Johns Hopkins McKusick-Nathans School of Medicine. Since 2011, Dr. Sobreira has also been part of the Baylor-Hopkins Center for Mendelian Genomics (BHCMG) team as part of the Phenotype Review Committee and Analysis Committee with a key role in variant discovery/confirmation.

SUMMARY:

Topic: PhenoDB: A Tool for Collection and Analysis of Phenotypic and Genomic Data

Speakers: Dr. Ada Hamosh and Dr. Nara Sobreira

Date: Wednesday, April 1, 2015

Time: 11 AM – 12 PM ET

You are invited to listen to Dr. Hamosh and Dr. Sobreira's presentation in Room 2W910-912 in the NCI Shady Grove Building on Medical Center Drive or via WebEx.

Presentation: A screen cast of the presentation will be available for viewing after the event on the NCI CBIIT Speaker Series YouTube Playlist . View the presentation slides.

About the NCI CBIIT Speaker Series:

The National Cancer Institute (NCI) Center for Biomedical Informatics and Information Technology (CBIIT) Speaker Series is a bi-weekly knowledge-sharing forum featuring both internal and external speakers on topics of interest to the biomedical informatics and research communities. For additional information, including past speaker series presentations, visit the CBIIT Speaker Series page.

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