January 21: Melissa Haendel, Ph.D., The Monarch Initiative: Semantic Phenotyping for Disease Diagnosis and Discovery

SYNOPSIS:

Elucidating disease and dysfunction requires understanding how genotypic variation relates to phenotypic outcomes. However, data integration and retrieval are key challenges as phenotype data is largely unstructured and is encoded in a variety of formats. In addition, we only know the functional consequences of mutation for less than 40% of the human coding genome. Use of semantically structured phenotype data from model organisms so as to be comparable to human can supplement the human data to aid disease discovery. The ontologies act not only as data standards within and across species, but provide the connection of anatomical form to phenotypic outcomes — thereby enabling deep anatomical analysis to persevere, grow, and shed new light on how biological systems function across scale. The use of cross-species anatomy and phenotype ontologies can be combined with exome analysis to support disease diagnosis. The integrated data across models provisions for the development of methods to ensure quality structured phenotyping for maximal analytic utility, as well as garnering an understanding for how different organisms provide different phenotypic insights into gene function. Semantically capturing interactions with environmental perturbants (such as exposures or drug treatments) and the change in phenotypic outcomes over time is challenging, but is increasingly relevant for rare disease, cancer, and other more common disorders, as we seek to stratify patients and better support precision medicine.

Session details...

BIO:

Dr. Melissa Haendel is the Director of the Ontology Development Group within the Oregon Health & Science University Library and the Department of Medical Informatics and Epidemiology and principal investigator of the Monarch Initiative. Dr. Haendel earned her Ph.D. in Neuroscience at the University of Wisconsin, Madison, and a B.A. in Chemistry from Reed College. She was a post-doctoral fellow at Oregon State University and the University of Oregon.

SUMMARY:

Topic: The Monarch Initiative: Semantic Phenotyping for Disease Diagnosis and Discovery

Speaker: Melissa Haendel, Ph.D.

Date: Wednesday, January 21, 2015

Time: 11 AM – 12 PM ET

You are invited to listen to Dr. Haendel's presentation in Room 2W908 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 . Dr. Haendel's presentation slides are available at http://www.slideshare.net/mhaendel/nci-cbiit-haendel12115.

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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