

# \*\*\* Scientific Knowledge Discovery over Big Data \*\*\*

1pm- 3pm, November 17<sup>th</sup>, 2016  
Marriott Marquis San Diego Marina  
333 W Harbor Drive, San Diego, CA 92101-7709  
Room: Rancho Santa Fe 3

Genome-wide association studies (GWAS) of the human brain, represent the examination of a complete set of genetic variants in different individuals to determine if any variant is associated with a structural or functional neurological trait. On the other hand, with the recently increased availability of detailed phenotypic data from electronic health records and epidemiological studies, the impact of one or more genetic variants on the phenome is starting to be characterized both in clinical and population-based settings using phenome-wide association studies (PheWAS). These studies reveal a number of challenges that will need to be overcome to unlock the full potential of PheWAS for the characterization of the complex human genome-phenome relationship. The Big Data for Discovery Science Center (BDDS; <http://bd2k.ini.usc.edu>) - comprised of leading experts in biomedical imaging, genetics, proteomics, and computer science - is taking focused an "-ome to home" approach toward streamlining big data management, aggregation, manipulation, integration, and the modeling of biological systems across spatial and temporal scales. In this SFN Satellite Symposium, featuring members of the BDDS team, we will provide a series of presentations/tutorials on the development of modern data processing workflows to accommodate the "big data" associated with GWAS and PheWAS brain imaging studies, illustrate end-to-end applications in the identification of neuroimaging-related biomarkers, as well as full-scale neuroimaging GWAS and PheWAS solutions.

The agenda for the symposium is as follows:

## Introduction to the BDDS Project and its Tools

**(15 mins):** *The Big Data for Discovery Science Center (BDDS) is a unique effort focused on the user experience with big data. Tools that are not only enabling but intuitive and adaptive are being created to directly answer these needs.*



*John Van Horn, Ph.D.*

**LONI Pipeline Fundamentals (30 mins):** *Here I will provide a presentation on the basics of how to develop and deploy "big data" processing workflows using the LONI Pipeline environment and its connections to cloud computing resources. LONI Pipeline forms the basis for many of the GWAS/PheWAS analyses attendees will see in the following lectures.*



*Sam Hobel*

**Mining Neuroimaging "big data" (30 mins):** *This presentation will demonstrate a statistical learning approach to explore neuroimaging "big data" in order to derive an association of interest. We will discuss some of the limitations of conventional regression techniques in big data analysis, and will show how statistical learning techniques provide increased inferential power.*



*Farshid Sepehrband, Ph.D.*

**GWAS Analyses (30 mins):** *GWAS of Quantitative Traits: A Semi-Automated Approach.* We demonstrate the use of state-of-the-art "big data" genomic and neuroimaging workflow tools in reducing the mining of quantitative trait loci to three simple steps. This approach is particularly conducive to "reproducible science" and the flexible visualization of association results.



*William Matloff*

**PheWAS Analyses (30 mins):** In this presentation, we will introduce a big data discovery framework for neuroimaging PheWAS, and provide an example of using the approach to identify which brain phenotypes out of 2,000,000's are influenced by a specific genetic factor. Such analyses provide unique insights into the role of specific genes on brain-specific phenotypes.



*Lu Zhao, Ph.D.*

**Interactive Q&A with all speakers**

**Hands-On Session (30 Mins)**

**Conclusion of Symposium**

*All Presenters*

This symposium is ideally suited for attendance by the neuroscientist, data scientist, and/or informatician interested in large-scale data processing, modern analytic methods, and their utility for novel discovery. All software tools and workflows for GWAS and PheWAS analytics will be available for download from the BDDS website and only available to those who attend this exclusive event. Supported by NIH P41 EB015922 and U54 EB020406 awards.