

# Genetics

Thursday, Jun 21: 10:30 AM - 11:45 AM

Oral Sessions

Thursday - Oral Session

## Presentations

### **1551: The ENIGMA Cortical GWAS Collaboration identifies 81 genetic loci influencing cortical structure.**

10:30 AM - 10:42 AM

In the last two decades, computational brain mapping approaches, together with behavioral genetics methods, have shown that the structure of the human neocortex is heritable [1]. Yet, relatively few studies have sought to identify specific genetic loci that influence cortical structure. Genetic loci that influence the brain's gray matter (GM) structure may also contribute to psychiatric disease risk, offering a more mechanistic understanding of these genetic risks. In addition, understanding the way in which genetic variants influence healthy brain structure provides an important baseline for characterizing the effects of genetic loci on brain disorders. While some disease differences can be detected in imaging studies of a few hundred subjects, these sample sizes are insufficient to discover the small effects of common genetic variants on brain structure. To overcome sample size limitations, over 200 researchers worldwide have performed a large-scale genome-wide association study (GWAS) of cortical measures, as part of the Enhancing Neuroimaging Genetics through Meta Analysis (ENIGMA) Consortium, to uncover the first robust set of genetic loci to influence cortical GM surface area and thickness.

Presenter

*Neda Jahanshad*, University of Southern California

### **1556: Genetics of brain structure and function: 3,144 GWAS from UK Biobank data**

10:42 AM - 10:54 AM

Joint analysis of both genetic and brain imaging UK Biobank datasets presents a unique opportunity for uncovering the genetic bases of brain structure and function, including genetic factors relating to brain development, ageing and disease. We carried out genome-wide association studies of 3,144 distinct brain imaging derived phenotypes from UK Biobank data.

Presenter

*Fidel Alfaró Almagro*, FMRIB Centre, WIN, University of Oxford

### **1092: Rare genetic events in sporadic Alzheimer's disease: a network propagation approach.**

10:54 AM - 11:06 AM

The discovery of rare genetic mutations in the APP, PSEN1, and PSEN2 that cause familial Alzheimer's disease (AD) has advanced our understanding of disease pathogenesis. There is much interest in investigating rare genetic variants that might increase risk for, or influence the phenotype of, sporadic AD. This remains challenging due to the very large sample sizes required and the costs of performing whole-genome sequencing (WGS) in these cohorts. Early successes have included the discovery of TREM2 (Guerreiro et al. 2013). Studies such as the Alzheimer's Disease Sequencing Project (Bis et al. 2017) continue to address this challenge. Here, we present a method to investigate the effect of rare variants in moderately sized sporadic AD cohorts, such as ADNI. Our approach is based on signal diffusion on a gene-interaction network, inspired by its successful application to cancer subtyping (Hofree et al. 2013). We seek to develop an integrative approach to guide gene-based association testing with the knowledge of tissue-specific interaction networks.

Presenter

*Marzia Antonella Scelsi*, University College London

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## **1564: Microstructural Heritability of the Corpus Callosum in Human**

11:06 AM - 11:18 AM

The corpus callosum (CC) is the largest commissure fiber in human brain and plays a fundamental role in interhemispheric information integration and cerebral lateralization [sup]1[/sup]. There have been a few MRI studies showing genetic effects on multiple CC traits (e.g. size, volume and microstructural properties) [sup]2[/sup]. To date, however, it remains scarce to study how genetic factors influence biologically specific measures of the CC, e.g., neurite density. To address this, we here employed the large-scale diffusion MRI datasets of Human Connectome Project (HCP) twins, and applied the AE model to quantify the heritability of multiple diffusion tensor imaging (DTI) and neurite orientation dispersion and density imaging (NODDI) [sup]3[/sup] measures.

Presenter

*Chenxi Zhao*, Beijing Normal University

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## **2457: Studying Genetic Impact on Resting State Connectivity Using Twin Brains**

11:18 AM - 11:30 AM

It is well established that monozygotic twins have identical genomes as opposed to dizygotic twins or siblings, implying that the only differences observed in their characteristics should be due to the environment, and not genetics[<sup>1</sup>]. Given the rise of interest in using resting state functional MRI as a tool to characterize a person[<sup>2</sup>], comparing the resting state connectivity patterns between the monozygotic and dizygotic twins may prove to be noteworthy in denoting the significance of the genetic impact exclusively on the functional brain.

Presenter

*Arman Kulkarni*, University of Wisconsin-Madison

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## **2573: A Multivariate Brain Atlas of Genetic Depression Risk**

11:30 AM - 11:42 AM

Traditional Imaging Genetics studies focus on uncovering potential relationships between a genetic variation (e.g. a Single Nucleotide Polymorphism; SNP) and whole-brain voxel values (1). While this line of investigation has provided numerous valuable insights, it disregards the fundamental, widely-accepted believe of geneticists and neuroscientists that no single genetic variation exerts substantial effects with regard to any psychiatric disorders, but that disorders such as for example Major Depressive Disorder (MDD) are determined by a multitude of genetic variations (2). In line with this notion, a recent large-scale study incorporating N=130,664 MDD cases and 330,470 controls identified 44 risk variants for MDD significant in a genome-wide association study (GWAS; (3)).

Presenter

*Tim Hahn*, University of Muenster

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