deep genomics

Donnelly Centre for Cellular + Biomolecular Research
UNIVERSITY OF TORONTO

"Scaling up Genetic Analysis"

Co-hosted by CCBR and Deep Genomics

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C. David Naylor Building
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Toronto, Ontario, M5S 3H2

ABSTRACT Sequencing technology determines the need for genome analysis tools that meet the challenges of scale. I will describe our efforts to develop a software package, hail, that uses spark and scala in a distributed model of computing to achieve large scale genome analysis and quality control. We can perform primary quality control analyses on whole genome sequencing datasets of ~5,000 individuals in under an hour. Using hail, we have performed analyses of education attainment on a sample of over 14,000 individuals, identifying a clear role of ultra-rare disruptive mutations. We further explored this class of variation across a wide range of traits and demonstrate that neuropsychiatric traits appear to have a directional burden effect in contrast to later onset systemic disease.

BIO Benjamin Neale is an assistant professor in the Analytic and Translational Genetics Unit at Massachusetts General Hospital (MGH), assistant professor in medicine at Harvard Medical School (HMS), and an associated researcher at the Broad Institute. Neale earned his Ph.D. in human genetics from King's College in London, UK, and completed his postdoctoral training in Daly's laboratory at Massachusetts General Hospital.

Neale is strongly committed to gaining insights into the genetics of common, complex human diseases. He has analyzed genetic data from large-scale studies of patients with ADHD, autism, age-related macular degeneration, type 2 diabetes, and metabolic disorders. Neale also analyzed data from the first ADHD genome-wide association study (GWAS) meta-analysis. Neale contributed to the development of software tools such as PLINK, one of the most frequently used packages for GWAS analysis. Neale is an active member of the broader Psychiatric GWAS Consortium analysis committee. Neale also led the design of the exome chip, a genotyping array that captures rare coding variation in a cost-effective manner.