



McGill

Department of
**Epidemiology, Biostatistics
and Occupational Health**

Biostatistics Seminars

Fall 2017

SPECIAL SEMINAR



Andriy Derkach, PhD

Postdoctoral Fellow
National Cancer Institute
Biostatistics Branch

Simplified Power Calculations for Genetic Association Studies

Wednesday, November 22, 2017

2:30 pm – 3:30 pm

Purvis Hall, 1020 Pine Ave. West, Room 25

ALL ARE WELCOME

Abstract:

Genome-wide association studies are now shifting focus from an analysis of common to uncommon and rare variants with an anticipation to explain additional variation in complex traits. As power for association testing for individual rare variants may often be low, various aggregate level association tests have been proposed to detect genetic loci that may contain clusters of causal variants. We showed that these methods can be divided into two classes: tests based on linear and composite statistics (e.g. variance-component tests). Typically power calculations for such tests require specification of a large number of parameters, including effect sizes and allele frequencies of individual markers, making them difficult to use in practice. In this presentation, we approximate power of linear and quadratic tests to varying degree of accuracy using a smaller number of key parameters, including the total genetic variance explained by multiple variants within a locus. Using the simplified power calculation methods, we then develop a mathematical framework to obtain bounds on the genetic architecture of an underlying trait given results from a genome-wide study. By using proposed framework, we observe important implications for lack or a limited number of findings in many currently reported studies. Finally, we provide insights into the required quality of annotation/functional information for identification of likely causal variants to make meaningful improvement in power of subsequent association tests.

....1/2



McGill

Department of
**Epidemiology, Biostatistics
and Occupational Health**

Biostatistics Seminars

Fall 2017

Bio:

Andriy Derkach is currently conducting his post-doctoral research in Biostatistics Branch of Division of Cancer Epidemiology and Genetics at the National Cancer Institute. His doctoral and post-doctoral training have focused on statistical methods that integrate high dimensional molecular and genetics data within or between studies to improve probability of detecting associations with disease. He received his doctoral training at University of Toronto, where he worked on developing statistical tests and designs for association studies with rare variants. As post-doctoral fellow, he has been involved in studies of continuous biomarkers, such as metabolite levels that reflect the effects of endogenous, environmental and genetic factors. Currently, he is interested in detecting subsets of biomarkers mediating the relationship between a known risk factor and cancer.

....2/2