

**STATISTICAL METHODS FOR GENETICS & GENOMICS  
- RESEARCH SEMINAR AND JOURNAL CLUB  
Details 2015-2016**

October 16: 12 noon – Seminar – Reports from IGES/ASHG

October 23: 12 noon – Journal Club - Andrew Paterson, SickKids  
Reading: *The UK10K Consortium*.  
The UK10K project identifies rare variants in health and disease.  
NATURE (2015) doi:10.1038/nature14962 Early access.  
<http://www.ncbi.nlm.nih.gov/pubmed/26367797>  
<http://www.nature.com/nature/journal/vaop/ncurrent/full/nature14962.html>

\*\*\*\*\*

November 13 12 noon – Seminar/Journal Club - Sarah Gagliano, CAMH  
Topic: Machine Learning applied to genomics  
Reading: "Machine learning applications in genetics & genomics", Nature Reviews Genetics 2015.  
<http://www.nature.com.myaccess.library.utoronto.ca/nrg/journal/v16/n6/full/nrg3920.html>

November 20 12 noon – Journal Club - Shelley Bull, Lunenfeld-Tanenbaum  
Topic: Association analysis of SNPs, gene expression, and disease  
Reading: Gamazon et al. Nature Genetics 2015; 47:1091-1098.  
A gene-based association method for mapping traits using reference transcriptome data  
<http://www.nature.com.myaccess.library.utoronto.ca/ng/journal/v47/n9/full/ng.3367.html>

November 27 12 noon – Journal Club - Wei Deng, Statistical Science  
Topic: Use of LD score regression to differentiate confounding biases from polygenicity in GWAS  
Reading: *Bulik-Sullivan, Loh, Finucane et al. Nat Genet. 2015;47:291–295.*  
<http://www.ncbi.nlm.nih.gov/pubmed/25642630>  
<http://www.nature.com.myaccess.library.utoronto.ca/ng/journal/v47/n3/full/ng.3211.html>

\*\*\*\*\*

December 4 12 noon – Research Seminar - Andrew Paterson, SickKids  
Topic: GWAS for a novel tissue measure leads to identification of an environmental factor with massive effect  
Background Reading: *Eny et al (2014) Diabetologia, 57(8), 1623-1634.*  
<http://link.springer.com/article/10.1007/s00125-014-3286-9>  
*Eny et al (2015) Diabetes Technology & Therapeutics, 17(10), 726-734.*  
<http://online.liebertpub.com/doi/10.1089/dia.2015.0017>

December 9 Wednesday 1-2 pm Special Seminar at SickKids (PGCRL 2A/2B)

**SICKKIDS CENTRE FOR BRAIN & MENTAL HEALTH  
& CHILD HEALTH EVALUATIVE SCIENCES**

**Presents** 'Reproducibility, Replication and Scientific Evidence'

Speaker: Dr. Veronica J. Vieland, PhD

Battelle Chair in Quantitative and Computational Biology Director;

Vice President for Computational Research, Battelle Center for Mathematical Medicine;

Professor of Pediatrics and Statistics,

The Research Institute at Nationwide Children's Hospital & The Ohio State University

Open Science Collaboration (2015) Estimating the reproducibility of psychological science.

Science. 2015 Aug 28;349(6251):aac4716. doi: 10.1126/science.aac4716.  
<http://www.sciencemag.org.myaccess.library.utoronto.ca/content/349/6251/aac4716.full?sid=a9d960b5-e852-4029-a650-06c1bfca1035>

\*\*\*\*\*

January 15 10 am – Seminar/Journal Club - Marie-Pierre Dubé, Montreal Heart Institute & David Henry, ICES/DLSPH

Background Reading: *Schwartz et al (2012) N Engl J Med. 367(22):2089-99.*  
Effects of dalcetrapib in patients with a recent acute coronary syndrome.

<http://www.ncbi.nlm.nih.gov/pubmed/23126252>

*Tardif et al (2015) Circ Cardiovasc Genet.8(2):372-82.*

Pharmacogenomic determinants of the cardiovascular effects of dalcetrapib

<http://www.ncbi.nlm.nih.gov/pubmed/25583994>

January 22 10 am – Journal Club - Linda Kachuri, Epidemiology, DLSPH

Topic: Mendelian Randomization Analysis

Reading: *Evans & Davey Smith (2015) Annu. Rev. Genomics Hum. Genet. 16:327-350*

Mendelian Randomization: New Applications in the coming age of Hypothesis-Free Causality

<http://www.annualreviews.org.myaccess.library.utoronto.ca/doi/full/10.1146/annurev-genom-090314-050016>

January 29 10 am – Seminar/Journal Club - Gord Fehringer, Lunenfeld-Tanenbaum

Topic: Subset-based Meta-Analysis

Background Reading: *Bhattacharjee et al (2012) Amer J Hum Genet. 90(5):821*

A subset-based approach improves power and interpretation for the combined analysis of genetic association studies of heterogeneous traits.

<http://www.sciencedirect.com/science/article/pii/S0002929712001590>

<http://www.ncbi.nlm.nih.gov/pubmed/2256009>

\*\*\*\*\*

February 5 12 noon - International Speaker Seminar

Guest: Guillaume Paré, McMaster University

For details, see: <http://www.stage.utoronto.ca/home/iss>

Talk Title: A Method to Estimate the Contribution of Regional Genetic Associations to Complex Traits from Summary Association Statistics

February 11 10 am – Seminar/Journal Club - Dave Soave, Biostatistics, DLSPH

Topic: Testing Calibration of Survival Models at Extremes of Event Risk

Background Reading: Song M, Kraft P, Joshi AD, Barrdahl M, Chatterjee N.

Testing calibration of risk models at extremes of disease risk.

Biostatistics. 2015 Jan;16(1):143-54. doi: 10.1093/biostatistics/kxu034. Epub 2014 Jul 14.

<http://biostatistics.oxfordjournals.org.myaccess.library.utoronto.ca/content/16/1/143.abstract>

February 26 10 am – Journal Club - Angelo Canty, Math & Stats, McMaster

Topic: Meta-analysis methods for rare variants

Reading: Tang & Lin (2015). AJHG 97, 35-53

Meta-analysis for Discovering Rare-Variant Associations:

Statistical Methods and Software Programs.

<http://www.sciencedirect.com/science/article/pii/S000292971500186X>

<http://www.ncbi.nlm.nih.gov/pubmed/26094574>

\*\*\*\*\*

March 18 10 am – Journal Club - Joseph Jamnik, Nutritional Sciences

Topic: Gene-environment interaction in GWAS

Reading: Boonstra *et al* (2016). *Am J Epidemiol*  
Tests for Gene-Environment Interactions and Joint Effects with Exposure Misclassification.  
<http://www.ncbi.nlm.nih.gov/pubmed/26755675>  
<https://aje-oxfordjournals-org.myaccess.library.utoronto.ca/content/183/3/237.full>

\*\*\*\*\*

April 22 10 am – Journal Club - Andrew Paterson, Sick Kids  
Reading: Exome Aggregation Consortium (ExAC) *et al* (2015)  
Analysis of protein-coding genetic variation in 60,706 humans  
<http://biorxiv.org/content/early/2015/10/30/030338>  
there's also a supplement  
<http://biorxiv.org/content/biorxiv/early/2015/10/30/030338.full.pdf>  
<http://biorxiv.org/content/biorxiv/suppl/2015/10/30/030338.DC1/030338-1.pdf>

April 29 10 am – Journal Club - Laurent Briollais, Lunenfeld-Tanenbaum Research Institute  
Topic: Implementing and Evaluating Genetic Risk-Stratified Screening Strategies for Common Cancers  
Primary Reading: Yen *et al*, *The Prostate* 75(8), 825-835 (2015)  
Prostate cancer screening using risk stratification based on a multi-state model of genetic variants  
<http://onlinelibrary.wiley.com.myaccess.library.utoronto.ca/doi/10.1002/pros.22964/full>  
<http://www.ncbi.nlm.nih.gov/pubmed/25683204>  
Other Reading: Burton *et al*, *Nature Genetics* 45,349–351 (2013)  
Public health implications from COGS & potential for risk stratification and screening  
<http://www.nature.com.myaccess.library.utoronto.ca/ng/journal/v45/n4/pdf/ng.2582.pdf>  
<http://www.nature.com.myaccess.library.utoronto.ca/icogs/>

\*\*\*\*\*

May 13 10 am – Seminar – Jerry Lawless, University of Waterloo  
Topic: Cohort sampling studies of survival time outcomes in genetic epidemiology  
Reading: Kulathinal *et al*, *Case-cohort design in practice – experiences from the MORGAM Project*  
*Epidemiologic Perspectives and Innovations* 4.15 (December 2007): p15  
<http://epi-perspectives.biomedcentral.com/articles/10.1186/1742-5573-4-15>  
<http://www.ncbi.nlm.nih.gov.myaccess.library.utoronto.ca/pmc/articles/PMC2216006/>  
Breslow *et al*, *Using the Whole Cohort in the Analysis of Case-Cohort Data*  
*American Journal of Epidemiology* (8 April 2009), 169(11), 1398-1405  
<http://aje.oxfordjournals.org.myaccess.library.utoronto.ca/content/169/11/1398.full>  
Zeng & Lin, *Efficient Estimation of Semiparametric Transformation Models for Two-Phase Cohort Studies*, *JASA* (March 2014) 109(505), 371-383  
<http://www.tandfonline.com.myaccess.library.utoronto.ca/doi/abs/10.1080/01621459.2013.842172>

\*\*\*\*\*

June 3 12 noon – STAGE International Speaker Seminar  
Guest Speaker: Shili Lin, Ohio State University  
For details, see: <http://www.stage.utoronto.ca/home/iss>  
Talk Title: Addressing the Correlated Feature in Sequencing-Based DNA Methylation Data for Detection of Differentially Methylated Regions

June 17 10 am – Seminar - Philip Awadalla, Ontario Institute for Cancer Research  
Title: Tools for studying Gene x Environmental Interactions on Phenotypes  
- Diving into the Environment Black Box  
Background Reading: Recombination affects accumulation of damaging and disease-associated mutations in human populations.  
Hussin JG, Hodgkinson A, Idaghmour Y, Grenier JC, Goulet JP, Gbeha E, Hip-Ki E, Awadalla P.  
*Nat Genet.* 2015 Apr;47(4):400-4. doi: 10.1038/ng.3216. Epub 2015 Feb 16.

<http://www.nature.com.myaccess.library.utoronto.ca/ng/journal/v47/n4/full/ng.3216.html>  
Exploiting gene expression variation to capture gene-environment interactions for disease..  
Idaghdour Y, Awadalla P. Front Genet. 2013 May 31;3:228. doi: 10.3389/fgene.2012.00228  
<http://journal.frontiersin.org/article/10.3389/fgene.2012.00228/full>

June 24

10 am – Seminar - Shelley Bull, Lunenfeld-Tanenbaum Research Institute

Topic: Regional testing of SNP sets in GWAS

Background Reading: Comparisons of multi-marker association methods to detect association between a candidate region and disease.

Ballard DH, Cho J, Zhao H. 2010 Genet Epidemiol 34:201-212. doi: 10.1002/gepi.20448.

<http://www.ncbi.nlm.nih.gov/pubmed/19810024>

Clique-based clustering of correlated SNPs in a gene can improve performance of gene-based multi-bin linear combination test

Yoo YJ, Kim SA, Bull SB. 2015. BioMed Research International, Vol 2015. Article ID 852341

<http://www.ncbi.nlm.nih.gov/pubmed/26346579>

\*\*\*\*\*