Course Syllabus - CHL 5228H Y 2023-2024

STATISTICAL METHODS FOR GENETICS & GENOMICS
- RESEARCH SEMINAR AND JOURNAL CLUB

TIME and PLACE:
    Fall term 10am – 12noon Friday (In Person)
        Room: SS 2101, Sidney Smith Hall – 100 St. George Street
    Winter term 10am - 12noon Friday (in Person)
        Room: TBA
      Seminar: 1 hour; Small Group Discussion: 1 hour.
    http://www.dlsph.utoronto.ca/students/current-students/timetables/
    https://www.dlsph.utoronto.ca/course/statistical-methods-for-genetics-genomics-research-seminar-and-journal-club/

Co-INSTRUCTORS:

Shelley Bull	Andrew Paterson
Professor, DLSPH	Professor, DLSPH
Senior Scientist,	Senior Scientist,
Lunenfeld-Tanenbaum Research Institute	Hospital for Sick Children Research
60 Murray Street, Box 18	PGCRL 686 Bay Street,
Room 5-226	Room 12.9710,
Email: bull@lunenfeld.ca	Email: andrew.paterson@sickkids.ca

Acknowledgement of Territory: *"We wish to acknowledge this land on which the University of Toronto operates. For thousands of years it has been the traditional land of the Huron-Wendat, the Seneca, and the Mississaugas of the Credit. Today, this meeting place is still the home to many Indigenous people from across Turtle Island and we are grateful to have the opportunity to work on this land."*

A. COURSE DESCRIPTION

Goals and Objectives:

To understand current developments in statistical genetic/genomic methods and current analytic issues in genetic epidemiology.

To become familiar with sources of methodology literature for the design and analysis of investigations in statistical genetics, statistical genomics, and genetic epidemiology.

To develop critical evaluation skills for underlying theory and/or applications of current study designs and statistical analysis methods.

To develop skills in communication and presentation in an inter-disciplinary setting.

Audience: Senior Graduate students in Biostatistics, Epidemiology or Statistics

Pre-requisites: Biostatistics/Statistics coursework at the graduate level
        and instructor permission
Pre/Co-requisite: CHL5224H or STA480/2080 or equivalent (with permission of the course instructors)

For graduate students/post-doctoral fellows interested in registering in the course for credit/audit, please send an email to both co-instructors and include a brief description of your program, background and pre/co-requisite status, and purpose in registering in the course.

Special features about course delivery:

One hour Research Seminar/Journal Club session, held 2-3 times per month, September through April with faculty participation.
    Topics from previous years: https://stage.utoronto.ca/smgg/

Seminar sessions will be followed by one hour small-group discussion for registered students and other participants, with faculty discussion leaders

Co-ordinated with the monthly <mark>International Speaker Seminar Series</mark> (ISSS) organized as part of the STAGE Training Program in Advanced Genetic Epidemiology.
https://canssiontario.utoronto.ca/events/stage-isss/
ISSS held Friday at 12 noon usually followed by Informal post seminar Q&A with guest speaker. There will be opportunities for a guest speaker-trainee group meeting.

Group size for discussion: 5-10 trainees (PhD students, post-doctoral fellows).

Faculty Discussion leaders:
Shelley Bull, Dalla Lana School of Public Health & Lunenfeld-Tanenbaum Research Institute
Andrew Paterson, Dalla Lana School of Public Health & SickKids Research Institute

Academic deadlines:
Deadline for scheduling student presentations: 24 November 2023.
Deadline to submit a one page paper outline: no later than 9 February 2024.
Due date for the Final paper: 5 April 2024.

B. ASSIGNMENTS AND EVALUATION

Students are expected to:
   (1) attend Friday journal club sessions & seminars, including CANSSI STAGE ISSS
   (2) present in one journal club session, and
   (3) submit a short paper.

Pass/Fail according to
      Participation in Seminars and Discussion Groups (Fall 2023 & Winter 2024)
      Presentation (Late Fall 2023 or early Winter term 2024)
      Final paper (end of the Winter term 2024)

      A pass is required in all three components

Some guidelines for the paper

   Set up a meeting with course faculty to help formulate the research question that you want to consider in the paper, and discuss how to address it.
   Submit a one page outline no later than 9 February 2024.
   Final paper is due 5 April 2024.

Formatting and Length:

Any standard journal style is acceptable, see for example guidelines for authors in *Genetic Epidemiology*, *Statistics in Medicine*, *Amer J of Human Genetics.*
Please submit with 12 pt font, and 1.5 line spacing.
Length of 10-15 (max) manuscript pages, excluding references, tables and figures.

C. POLICY STATEMENTS

   1. **Respect for classmates:** The University of Toronto is committed to equity, human rights, and respect for diversity. All members of the learning environment in this course should strive to create an atmosphere of mutual respect where all members of our community can express themselves, engage with each other, and respect one another's differences. U of T does not condone discrimination or harassment against any persons or communities.

2. **Academic Integrity**: Students must adhere to the [Code of Behaviour on Academic Matters](#). **It is your responsibility to know what constitutes appropriate academic behaviour.** You are responsible for ensuring that you do not act in such a way that would constitute cheating, misrepresentation, or unfairness, including but not limited to, using unauthorized aids and assistance, personating another person, and committing plagiarism. For more information see [U of T Academic Integrity](#) website. Academic integrity includes understanding appropriate research and citation methods. If you are uncertain about this, please seek out additional information from the instructors or from other institutional resources including the following:

- This tip sheet provides clear and helpful information about appropriate academic citation: http://guides.library.utoronto.ca/citing
- This site offers a series of scenarios to help students understand how to prevent themselves from being subject to academic offence allegations https://www.utm.utoronto.ca/academic-integrity/students/scenarios
- Before handing in assignments students can also review this [academic integrity checklist](#) provided by the UofT Centre of Teaching Support & Innovation:
    - I have acknowledged the use of another's ideas with accurate citations.
    - If I used the words of another (e.g., author, instructor, information source), I have acknowledged this with quotation marks (or appropriate indentation) and proper citation.
    - When paraphrasing the work of others, I put the idea into my own words and did not just change a few words or rearrange the sentence structure
    - I have checked my work against my notes to be sure I have correctly referenced all direct quotes or borrowed ideas.
    - My references include only the sources used to complete this assignment.
    - This is the first time I have submitted this assignment (in whole or in part) for credit.
    - Any proofreading by another was limited to indicating areas of concern which I then corrected myself.
    - This is the final version of my assignment and not a draft.
    - I have kept my work to myself and did not share answers/content with others, unless otherwise directed by my instructor.
    - I understand the consequences of violating the University's Academic Integrity policies as outlined in the [Code of Behaviour on Academic Matters](#).

3. **Statement re use of generative AI tools:** Students may use artificial intelligence tools, including generative AI, in this course as learning aids or to help produce assignments. However, students are ultimately accountable for the work they submit. Students must submit, as an appendix with their assignments, any content produced by an artificial intelligence tool, and the prompt used to generate the content. Any content produced by an artificial intelligence tool must be cited appropriately. Many organizations that publish standard citation formats are now providing information on citing generative AI (e.g., MLA: https://style.mla.org/citing-generative-ai/). Students may choose to use generative artificial intelligence tools as they work through the assignments in this course; this use must be documented in an appendix for each assignment. The documentation should include what tool(s) were used, how they were used, and how the results from the AI were incorporated into the submitted work.

   July 2023: The School of Graduate Studies (SGS) has announced new [Guidance on the Appropriate Use of Generative Artificial Intelligence in Graduate Theses](#) which will be of interest to graduate students, supervisors, supervisory committee members, Graduate Chairs and Graduate Units. Example of guidance from peer-review journal: https://jamanetwork.com/journals/jama/fullarticle/2807956

4. **Accessibility**: Students with diverse learning styles and needs are welcome in this course. If you have a disability or health consideration that may require accommodations, please feel free to approach me/us and/or the Accessibility Services Office as soon as possible. The Accessibility Services staff are available by appointment to assess specific needs, provide referrals and arrange appropriate accommodations. The sooner you let them and me know your needs, the quicker we can assist you in achieving your learning goals in this course. For more information, or to register with Accessibility Services, please visit: http://studentlife.utoronto.ca/as.

D. SEMINAR SCHEDULE (subject to revision)

September 15   10 am – Information session for graduate students interested in registering in the course for credit/audit (Room SS 2101, Sidney Smith Hall – 100 St. George Street)

                    * September 20 – Last date to add a course *

September 22   10 am – Organizational Meeting re topics & themes for the Seminar/Journal Club this academic year
                    (In person Room TBA and Zoom)

September 29   10am – Seminar/Journal Club - TBA

October 6   10am – Trainee Discussion
                    Reading: Open problems in human trait genetics, *Genome Biol* 23, 131 (2022)
                    https://doi.org/10.1186/s13059-022-02697-9

October 13   12 noon – CANSSI STAGE International Speaker Seminar
            Speaker: Ron Do, Icahn School of Medicine at Mount Sinai, New York
            Charles Bronfman Professor, Personalized Medicine & Professor, Genetics and Genomic Sciences
                    https://canssiontario.utoronto.ca/event/stage-isss-ron-do/

October 20   10 am – Seminar/Journal Club - TBA
October 28   10 am – Seminar/Journal Club - TBA

November 3   No Seminar **ASHG November 1-4, IGES November 5-7 **

November 10   10 am – Seminar – Highlights from ASHG/IGES
November 17   10 am – Seminar/Journal Club. - TBA
November 24   10 am – Seminar/Journal Club - TBA

        * November 24 - Deadline to schedule course presentation in Winter term *

December 1   12 noon – CANSSI STAGE International Speaker Seminar
            Speaker: Eimear Kenny, Icahn School of Medicine at Mount Sinai, New York
                    Director, Institute for Genomic Health & Interim Chief, Division of Genomic Medicine
            Director, Center for Clinical Polygenic Risk & Professor of Medicine and Genetics

Population Genetics in An Era of Genomic Health
                    https://canssiontario.utoronto.ca/event/stage-isss-eimear-kenny/

        * last week of classes December 4 – 8 *

December 8   10 am – Seminar/Journal Club – TBA

********** **2024** ********** **(subject to revision)**

            * First week of classes January 8 -12

January 12   10 am – Research Seminar – TBA
January 19   10 am – Seminar/Journal Club –   TBA
January 26   10 am – Research Seminar – TBA

February 2   12 noon – CANSSI STAGE International Speaker Seminar - TBA
            Speaker: Michael Wu, Fred Hutchinson Cancer Research Center, University of Washington
                    Member, Public Health Sciences Division & Professor, Biostatistics Program
                    https://canssiontario.utoronto.ca/event/stage-isss-michael-wu/

February 9   10 am – <mark>Journal Club</mark> – TBA

* February 9 - Deadline to submit paper outline *

February 16   10 am – <mark>Journal Club</mark> – TBA

* February 20 - Final date to drop full-year course *

February 23   *No seminar* – Winter Break (Reading) Week

March 1   12 noon – <mark>CANSSI STAGE International Speaker Seminar</mark>
      Speaker: Genevieve Wojcik, Johns Hopkins Bloomberg School of Public Health
                              Assistant Professor of Epidemiology
                  https://canssiontario.utoronto.ca/event/stage-isss-genevieve-wojcik/

March 8   10 am – <mark>Seminar/Journal Club</mark> - TBA
March 15   10 am – <mark>Seminar/Journal Club</mark> - TBA
March 22   10 am – <mark>Seminar/Journal Club</mark> - TBA
March 29   10 am – No Seminar

** Good Friday holiday March 29 **
** Last week of classes – April 2 – 5 **

** April 5 - Due date for final student papers **

April 5   12 noon – <mark>CANSSI STAGE International Speaker Seminar</mark>
      Speaker: Catherine Tcheandjieu, Gladstone Institutes & UC San Francisco
                              Investigator & Assistant Professor, Dept of Epidemiology & Biostatistics
                  https://stage.utoronto.ca/isss/

May 3   12 noon – <mark>CANSSI STAGE International Speaker Seminar</mark>
      Speaker: Li Hsu, Fred Hutchinson Cancer Research Center, University of Washington
                              Member, Public Health Sciences Division & Affiliate Professor, Biostatistics
                  https://stage.utoronto.ca/events/stage-isss-li-hsu/

June 7   12 noon – <mark>CANSSI STAGE International Speaker Seminar</mark>
      Speaker: Jonathan Marchini, Regeneron Genetics Center
                        Head, Statistical Genomics and Machine Learning
                  https://stage.utoronto.ca/events/stage-isss-jonathan-marchini/