



BINF*6110 Genomic Methods for Bioinformatics

Winter 2023

Section(s): C01

College of Biological Science

Credit Weight: 0.50

Version 1.00 - January 06, 2023

1 Course Details

1.1 Calendar Description

This course provides an introduction to current and emerging methods used to generate genomic data analyzed in bioinformatics. This may include techniques for DNA sequencing as well as transcriptome, proteome and metabolome analysis. The objective is to develop an appreciation for the challenges of producing data.

Restrictions: Restricted to Bioinformatics students.

1.2 Course Description

Genomic Methods for Bioinformatics (BINF*6110, winter 2022) will focus on the analysis of high-throughput sequence data that students can expect to encounter in current bioinformatics applications. We will focus on four topics: 1) types of genomic data and their application domains; 2) alignment and assembly, including choice of reference genome; 3) detection and filtering of genetic variants; 4) advanced analyses based on these variants. This course will involve hands-on exercises in a high performance Unix computing environment, as well as analysis and data visualization in R; previous experience in both Unix and R is an asset.

1.3 Timetable

Dates: Jan. 10, 2023 - April 4, 2023

Time: Tuesday/Thursday 11:30-12:50

Location: SSC 1306

1.4 Final Exam

There will not be a final exam for this course.

2 Instructional Support

2.1 Instructional Support Team

Instructor:	Elizabeth Mandeville
Email:	emandevi@uoguelph.ca
Telephone:	+1-519-824-4120 x52843
Office:	SSC 1454
Office Hours:	By appointment

3 Learning Resources

In this course we will not have a specific textbook, but will instead rely on a variety of resources, including online tutorials, software documentation, and the primary literature. Readings and links will be posted to the course website as they are assigned.

3.1 Additional Resources

Bioinformatics Data Skills (Textbook)

<https://ebookcentral.proquest.com/lib/uoguelph/detail.action?docID=3564550>

Bioinformatics Data Skills by Vince Buffalo is a valuable resource to complement the learning in this class. Note that this book is available as an electronic resource from the University of Guelph library, so you do not need to purchase a paper copy unless you prefer that format. Note that this book is from 2015, which is a long time ago in bioinformatics (sequencing and analysis change fast), but it is still relevant even if some syntax has changed. Code examples are also available at <https://github.com/vsbuffalo/bds-files/>

4 Learning Outcomes

4.1 Course Learning Outcomes

By the end of this course, you should be able to:

1. Describe currently available types of genomic data, and identify suitable applications and limitations
 2. Choose suitable genomic resources and software for bioinformatics analyses
 3. Work effectively in a Unix high-performance computing environment
 4. Communicate the results of bioinformatic analyses in written, oral, and visual format
-

5 Teaching and Learning Activities

This class will be a mixture of mini-lectures where we review content, and working sessions where students apply new concepts to the analysis of actual data. We will be working on an HPC (high performance computing) cluster for the majority of exercises in this class. Note that the HPC systems we will use have remote access through Unix SSH, so you will be able to access these systems through your laptop or lab computers - no special hardware needed. If you intend to use your own laptop, you must install a Unix/Linux Client on your laptop. Guidance and set-up assistance will be provided in the first week of the course. Additionally, all students are expected to have R installed on their laptops.

We will focus on four topics during this course: 1) Types of genomic data and their uses; 2) Genome assembly and alignment of sequence data to a reference; 3) Genetic variant identification and filtering; 4) Common analyses applied to genomic data.

6 Assessments

This course will be project-based, and will feature four major assignments (akin to problem sets or lab reports), each worth 20-25% of your grade for the course. Assignments will correspond to the four major sections of the course; more detailed instructions will be provided at least 2 weeks prior to the due date. Each assignment will involve a combination of written report, code, and figures.

We will also have a number of smaller assignments due in weeks when no larger assignment is due. Note that these small assignments will not be time consuming, and are intended to check your progress; they are also a tool for the instructor to understand which topics might need more attention. These assignments will cumulatively be worth 10% of your grade.

6.1 Assessment Details

Assignment 1: Due by 4 PM Friday, January 27, 2023 (20%)

Will focus on finding publicly available genomic resources and quantifying characteristics of reference genomes

Assignment 2: Due by 4 PM Friday, Feb 17 (25%)

Focus on genome assembly and alignment to reference genomes

Assignment 3: Due by 4 PM Friday, March 17 (25%)

Will focus on identification and filtering of genomic variants (SNPs and INDELS)

Assignment 4: Due by 4 PM Monday, April 3 (20%)

Will focus on an advanced topic of the student's choice. Final presentations in class April 4 and April 6

Small check-in assignments (10%)

Due weekly on Fridays (by 4 PM) in some weeks with no larger assignment. More information will be given in the week before each small assignment is due. These assignments are meant to serve as a check-in between instructor and students, and provide practice and reinforcement for new concepts.

7 Course Statements

7.1 Attendance

Attendance is strongly suggested for this course. All classes will include crucial hands-on problem solving experiences that can not easily be replaced outside of regular class sessions. However, lecture portions of the class will likely be recorded and available on Courselink for students who are unable to avoid missing a class session due to illness or personal circumstances. Students who are ill are encouraged to stay home and make use of recordings and posted notes, as well as opportunities to meet with the instructor virtually to review any questions you may have about new material.

7.2 Collaboration and group work

Assignments for this course are individual, but students may confer and seek help from one another to complete assignments. I encourage you to learn from one another as you work through new types of analyses. However, note that all code, text, and figures must be the individual work of the student submitting it, and that duplicate submissions (including identical code for reasonably complex tasks) will be treated as plagiarism and a violation of Academic Integrity standards (please see below).

7.3 Due dates

Assignments are due by the dates listed in the Course Outline. Late submissions for major assignments (without a pre-approved extension) will be marked down 10% for each day that they are late. If you are ill or cannot complete your assignments on time for any reason, please contact the instructor as soon as possible; accommodations are possible. For Small Check-in Assignments, each student may skip one assigned item without penalty, no questions asked and no excuse needed. Additionally, for major assignments (Projects 1-4), each student gets one no-questions-asked 48-hour extension without penalty.

Please know that I regard your health and safety as the top priority; I will work with you to help fulfill academic requirements. If you are unable to meet the requirements of the course for any reason, please contact me and we will work towards a solution.

8 College of Biological Science Statements

8.1 Wellness

If you are struggling with personal or health issues:

- Counselling Services offers individualized appointments to help students work through personal struggles that may be impacting their academic performance.
- Student Health Services is located on campus and is available to provide medical attention.
- For support related to stress and anxiety, besides Health Services and Counselling Services, Kathy Somers runs training workshops and one-on-one sessions related to stress management and high performance situations.
<http://www.selfregulationskills.ca/>

8.2 Personal information

Personal information is collected under the authority of the University of Guelph Act (1964), and in accordance with Ontario's Freedom of Information and Protection of Privacy Act (FIPPA) <http://www.e-laws.gov.on.ca/index.html>. This information is used by University officials in order to carry out their authorized academic and administrative responsibilities and also to establish a relationship for alumni and development purposes.

For more information regarding the Collection, Use and Disclosure of Personal Information policies please see the Undergraduate Calendar.
(<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/intro/index.shtml>)

8.3 Course Offering Information Disclaimer

Please note that course delivery format (face-to-face vs online) is subject to change up to the first-class day depending on requirements placed on the University and its employees by public health bodies, and local, provincial and federal governments. Any changes to course format prior to the first class will be posted on WebAdvisor/Student Planning as they become available.

9 University Statements

9.1 Email Communication

As per university regulations, all students are required to check their e-mail account regularly: e-mail is the official route of communication between the University and its students.

9.2 When You Cannot Meet a Course Requirement

When you find yourself unable to meet an in-course requirement because of illness or compassionate reasons please advise the course instructor (or designated person, such as a teaching assistant) in writing, with your name, id#, and e-mail contact. The grounds for Academic Consideration are detailed in the Undergraduate and Graduate Calendars.

Undergraduate Calendar - Academic Consideration and Appeals
<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-ac.shtml>

Graduate Calendar - Grounds for Academic Consideration

<https://www.uoguelph.ca/registrar/calendars/graduate/current/genreg/index.shtml>

Associate Diploma Calendar - Academic Consideration, Appeals and Petitions
<https://www.uoguelph.ca/registrar/calendars/diploma/current/index.shtml>

9.3 Drop Date

Students will have until the last day of classes to drop courses without academic penalty. The deadline to drop two-semester courses will be the last day of classes in the second semester. This applies to all students (undergraduate, graduate and diploma) except for Doctor of Veterinary Medicine and Associate Diploma in Veterinary Technology (conventional and alternative delivery) students. The regulations and procedures for course registration are available in their respective Academic Calendars.

Undergraduate Calendar - Dropping Courses
<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-drop.shtml>

Graduate Calendar - Registration Changes
<https://www.uoguelph.ca/registrar/calendars/graduate/current/genreg/genreg-reg-regchg.shtml>

Associate Diploma Calendar - Dropping Courses
<https://www.uoguelph.ca/registrar/calendars/diploma/current/c08/c08-drop.shtml>

9.4 Copies of Out-of-class Assignments

Keep paper and/or other reliable back-up copies of all out-of-class assignments: you may be asked to resubmit work at any time.

9.5 Accessibility

The University promotes the full participation of students who experience disabilities in their academic programs. To that end, the provision of academic accommodation is a shared responsibility between the University and the student.

When accommodations are needed, the student is required to first register with Student Accessibility Services (SAS). Documentation to substantiate the existence of a disability is required; however, interim accommodations may be possible while that process is underway.

Accommodations are available for both permanent and temporary disabilities. It should be noted that common illnesses such as a cold or the flu do not constitute a disability.

Use of the SAS Exam Centre requires students to make a booking at least 14 days in advance, and no later than November 1 (fall), March 1 (winter) or July 1 (summer). Similarly, new or changed accommodations for online quizzes, tests and exams must be approved at least a week ahead of time.

For Guelph students, information can be found on the SAS website
<https://www.uoguelph.ca/sas>

For Ridgetown students, information can be found on the Ridgetown SAS website
<https://www.ridgetownc.com/services/accessibilityservices.cfm>

9.6 Academic Integrity

The University of Guelph is committed to upholding the highest standards of academic integrity, and it is the responsibility of all members of the University community-faculty, staff, and students-to be aware of what constitutes academic misconduct and to do as much as possible to prevent academic offences from occurring. University of Guelph students have the responsibility of abiding by the University's policy on academic misconduct regardless of their location of study; faculty, staff, and students have the responsibility of supporting an environment that encourages academic integrity. Students need to remain aware that instructors have access to and the right to use electronic and other means of detection.

Please note: Whether or not a student intended to commit academic misconduct is not relevant for a finding of guilt. Hurried or careless submission of assignments does not excuse students from responsibility for verifying the academic integrity of their work before submitting it. Students who are in any doubt as to whether an action on their part could be construed as an academic offence should consult with a faculty member or faculty advisor.

Undergraduate Calendar - Academic Misconduct

<https://www.uoguelph.ca/registrar/calendars/undergraduate/current/c08/c08-amisconduct.shtml>

Graduate Calendar - Academic Misconduct

<https://www.uoguelph.ca/registrar/calendars/graduate/current/genreg/index.shtml>

9.7 Recording of Materials

Presentations that are made in relation to course work - including lectures - cannot be recorded or copied without the permission of the presenter, whether the instructor, a student, or guest lecturer. Material recorded with permission is restricted to use for that course unless further permission is granted.

9.8 Resources

The Academic Calendars are the source of information about the University of Guelph's procedures, policies, and regulations that apply to undergraduate, graduate, and diploma programs.

Academic Calendars

<https://www.uoguelph.ca/academics/calendars>

9.9 Disclaimer

Please note that the ongoing COVID-19 pandemic may necessitate a revision of the format of course offerings, changes in classroom protocols, and academic schedules. Any such

changes will be announced via CourseLink and/or class email.

This includes on-campus scheduling during the semester, mid-terms and final examination schedules. All University-wide decisions will be posted on the COVID-19 website (<https://news.uoguelph.ca/2019-novel-coronavirus-information/>) and circulated by email.

9.10 Illness

Medical notes will not normally be required for singular instances of academic consideration, although students may be required to provide supporting documentation for multiple missed assessments or when involving a large part of a course (e.g.. final exam or major assignment).

9.11 Covid-19 Safety Protocols

For information on current safety protocols, follow these links:

- <https://news.uoguelph.ca/return-to-campusess/how-u-of-g-is-preparing-for-your-safe-return/>
- <https://news.uoguelph.ca/return-to-campusess/spaces/#ClassroomSpaces>

Please note, these guidelines may be updated as required in response to evolving University, Public Health or government directives.
