Computer Science EN.601.449 / EN.601.649
Computational Genomics: Applied Comparative Genomics
Fall, 2022 (3 credits, EQ)

Instructor
Professor Michael Schatz, mschatz@cs.jhu.edu, http://schatz-lab.org
Office: Malone 323
Office hours: TBD; and by appointment

Teaching Assistant
Alex Sweeten, asweete1@jhu.edu
Office: Malone Hall
Office hours: TBD; and by appoinment

Meetings
Monday, Wednesday, 3:00pm – 4:15pm, Hodson 311
Recorded lectures can be accessed on the course Canvas site

Textbook
No textbooks are required.

Online Resources
The following online resources are essential:

- The course web site contains a schedule of topics, class notes, and assignment details: https://github.com/schatzlab/appliedgenomics2023

- The course Piazza site at https://piazza.com/class/llqpylue3c4v9/. This site will serve as our discussion site for the course. Please use Piazza to ask questions of the instructor, TA and fellow students.

Course Information

- The goal of this course is to study the leading computational and quantitative approaches for comparing and analyzing genomes starting from raw sequencing data. The course will focus on human genomics and human medical applications, but the techniques will be broadly applicable across the tree of life. The topics will include genome assembly & comparative genomics, variant identification & analysis, gene expression & regulation, personal genome analysis, and cancer genomics. The grading will be based on assignments, a midterm exam, class presentations, and a significant class project.
- **Prerequisites:** knowledge of the Unix operating system and programming expertise in a language such as R or Python. [Applications]

Course Goals

Upon successful completion of this course, you should be able to:

1. Understand the theoretical foundations for several of the most important genomic analysis tools
2. Have hands-on experience running several of the most important genomic tools
3. Perform novel research and analysis in computational biology

Course Topics

We will study the leading computational and quantitative approaches for comparing and analyzing genomes starting from raw sequencing data. The course will focus on human genomics and human medical applications, but the techniques will be broadly applicable across the tree of life. The topics will include genome assembly & comparative genomics, variant identification & analysis, gene expression & regulation, personal genome analysis, and cancer genomics. Please see the main course website for a more detailed schedule, which will be updated as the semester progresses

Course Expectations & Grading

Course grades will be based on assignments (typically running and analyzing existing tools and/or analyzing a dataset), an examination, and a class project, according to the proportions below. Each homework assignment will be assigned a point value; the overall homework assignment grade will be computed as your total points earned divided by the total achieved in the class.

- 30% - Assignments
- 30% - Midterm (Take home; See class schedule for dates)
- 40% - Class Project (Final report along with an in-class presentation and preliminary report)

All grades will be distributed via email. Please keep your own record of your grades so that you will know your standing in the course. Letter grades for the course will be assigned on a standard scale, subject to the instructor's evaluation of your overall class performance. Students are allowed a total of 96 hours to extend the deadline for assignments, but not the class project, without any penalty. No further extensions will be allowed without a doctor's note or a note from the university.

Assignment Logistics. The assignments and projects in this course will require you to execute command line programs, write code in the language of your choice, or carry out a calculation. You must write all code independently unless the assignment specifically states that you can work in groups. Assignments will be submitted via Gradescope.

Attendance. All students are generally expected to attend all meetings of this course, and actively participate in all course meetings. If you miss a class meeting for any reason, you

are responsible for material presented, and it is your responsibility to obtain any missed handouts or other materials. If you will be missing more than 1 class, please contact the instructor to discuss how to best review the missed materials.

Key Dates
See the schedule on the class website

Assignments & Readings
See the schedule on the class website

Ethics
The strength of the university depends on academic and personal integrity. In this course, you must be honest and truthful. Ethical violations include cheating on exams, plagiarism, reuse of assignments, improper use of the Internet and electronic devices, unauthorized collaboration, alteration of graded assignments, forgery and falsification, lying, facilitating academic dishonesty, and unfair competition.
[In addition, the specific ethics guidelines for this course are:
(1) (Insert unique rules here, such as your policy regarding collaboration on assignments or use of old exams/graded materials.)
(2) (etc.)]
Report any violations you witness to the instructor. You can also contact:

- For undergraduates: the director of student conduct (or designee) by calling the Office of Student Conduct at 410-516-2509 or via email at studentconduct@jhu.edu
- For KSAS Graduate Students; rseitz5@jh.edu
- For WSE Graduate Students: christinekavanagh@jhu.edu

Personal Wellbeing

- Because of ongoing COVID-19 concerns, special requirements may be in effect this term, and these may vary during the term. Please keep updated with these at the following sites:
  - University information: https://covidinfo.jhu.edu/
  - Whiting School of Engineering information: https://engineering.jhu.edu/covid-19/
- COVID-19 vaccination a required unless an exception has been granted by the university for health or religious reasons.
- If you are sick please notify me by email so that we can make appropriate accommodations should this affect your ability to attend class, complete assignments, or participate in assessments. The Student Health and Wellness Center is open and operational for primary care needs. If you would like to speak with a medical provider, please call 410-516-8270, and staff will determine an appropriate course of action. See also https://studentaffairs.jhu.edu/student-life/student-outreach-support/absences-from-class/illness-note-policy/
- Johns Hopkins University values diversity and inclusion. We are committed to providing welcoming, equitable, and accessible educational experiences for all

students. Students with disabilities (including those with psychological conditions, medical conditions, and temporary disabilities) can request accommodations for this course by providing an Accommodation Letter issued by Student Disability Services (SDS). Please request accommodations for this course as early as possible to provide time for effective communication and arrangements.

For further information or to start the process of requesting accommodations, please contact Student Disability Services at Homewood Campus, Shaffer Hall #101, call: 410-516-4720 and email: studentdisabilityservices@jhu.edu or visit the website https://studentaffairs.jhu.edu/disabilities/.

- If you are struggling with anxiety, stress, depression, or other mental health related concerns, please consider visiting the JHU Counseling Center. If you are concerned about a friend, please encourage that person to seek out their services. The Counseling Center is located at 3003 North Charles Street in Suite S-200 and can be reached at 410-516-8278 and online at http://studentaffairs.jhu.edu/counselingcenter/

- Student Outreach & Support helps students manage physical and mental health concerns, personal and family emergencies, financial issues, and other obstacles that may arise during their college experience. Students can self-refer or refer a friend who may need extra support or help getting connected to resources. To connect with SOS, please visit this website: https://studentaffairs.jhu.edu/student-life/student-outreach-support/ or email deanofstudents@jhu.edu, call 410-516-7857, or students can schedule to meet with a Case Manager by visiting the Student Outreach & Support website and filling out a referral form online.

Classroom Climate

I am committed to creating a classroom environment that values the diversity of experiences and perspectives that all students bring. Everyone here has the right to be treated with dignity and respect. I believe fostering an inclusive climate is important because research and my experience show that students who interact with peers who are different from themselves learn new things and experience tangible educational outcomes. Please join me in creating a welcoming and vibrant classroom climate. Note that you should expect to be challenged intellectually by me, the TAs, and your peers, and at times this may feel uncomfortable. Indeed, it can be helpful to be pushed sometimes in order to learn and grow. But at no time in this learning process should someone be singled out or treated unequally on the basis of any seen or unseen part of their identity.

If you ever have concerns in this course about harassment, discrimination, or any unequal treatment, or if you seek accommodations or resources, I invite you to share directly with me or the TAs. I promise that we will take your communication seriously and seek mutually acceptable resolutions and accommodations. Reporting will never impact your course grade. You may also share concerns with the department chair ([Chair's Name and Email]), the Director of Undergraduate Studies ([DUS Name and Email]), the WSE Assistant Dean for Diversity and Inclusion (Darlene Saporu, dsaporu@jhu.edu), the KSAS Assistant Dean for Diversity and Inclusion (Araceli Frias, afrias3@jhu.edu) or the

Office of Institutional Equity (oie@jhu.edu). In handling reports, people will protect your privacy as much as possible, but faculty and staff are required to officially report information for some cases (e.g., sexual harassment).

Family Accommodations Policy

You are welcome to bring a family member to class on occasional days when your responsibilities require it (for example, if emergency child care is unavailable, or for health needs of a relative). In fact, you may see my children in class on days when their school is closed. Please be sensitive to the classroom environment, and if your family member becomes uncomfortably disruptive, you may leave the classroom and return as needed.

Policy on Incomplete Grades:

The full policy is available here:
https://e-catalogue.jhu.edu/engineering/full-time-residential-programs/undergraduate-policies/academic-policies/grading-policies/
The following text is an excerpt:

1.   *A request for an Incomplete grade must be initiated by the student no later than the last day of classes via the Incomplete Grade Contract available in SIS*
2.   *The required elements on the Incomplete Grade Contract are listed below; all of these topics should be included in the conversation between the student and the instructor.*
- *The reason for the request for an incomplete grade*
- *A description of all outstanding work that must be completed*
- *Date the work is due from the student*
- *The reversion grade if the student does not complete any of the outstanding work*

3. Instructors are required to submit the new grade to the Office of the Homewood Registrar no later than 45 calendar days after the last day of classes. If the Incomplete grade is not resolved within 45 calendar days after the last day of classes, the Incomplete grade is automatically converted to the reversion grade.

The significant component of this policy is that there is an Incomplete Grade Contract available to students in SIS to request an incomplete grade. This is how all incomplete grades must be initiated. Another significant component is the timeline for completion of an incomplete grade for undergraduate students, which is **45 calendar days after the last day of classes**. For graduate students, the deadline is the end of the third week of classes of the following semester. See the full catalogue entry for considerations for students on academic probation and graduating students.

Deadlines for Adding, Dropping and Withdrawing from Courses
https://studentaffairs.jhu.edu/registrar/students/registration/

Full-semester classes:
Students may add a course up to **noon on September 8, 2023** (customized academic learning, formerly called independent academic work, such as research may be added until **October 8, 2023**). They may drop courses up until **October 8, 2023** provided they remain registered for a minimum of 12 credits. Between **October 9, 2023 and November 10, 2023,** a student may withdraw from a course with a W on their academic record. A record of the course will remain on the academic record with a W appearing in the grade column to indicate that the student registered and then withdrew from the course.

Term I classes:
Students may add a course up to **September 8, 2023.** They may drop courses until **September 17, 2023** provided they remain registered for a minimum of 12 credits. Between **September 18, 2023 and September 29, 2023,** a student may withdraw from a course with a W on their academic record. A record of the course will remain on the academic record with a W appearing in the grade column to indicate that the student registered and then withdrew from the course.

Term II classes:
Students may add a course up to **November 3, 2023.** They may drop courses up until **November 12, 2023** provided they remain registered for a minimum of 12 credits. Between **November 13, 2023 and December 1, 2023,** a student may withdraw from a course with a W on their academic record. A record of the course will remain on the academic record with a W appearing in the grade column to indicate that the student registered and then withdrew from the course.

Please see the Registrar's website for relevant deadlines for HEART/HEROIC and SOUL courses.

For more information on these and other academic policies, see
https://e-catalogue.jhu.edu/engineering/full-time-residential-programs/undergraduate-policies/academic-policies/grading-policies/