

# 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**

Bohan Ni, bni1@jhu.edu Office: Malone Hall

Office hours: TBD; and by appointment

#### Meetings

Monday, Wednesday, 1:30-2:45pm, Gilman 17

#### **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: <a href="https://github.com/schatzlab/appliedgenomics2022">https://github.com/schatzlab/appliedgenomics2022</a>
- The course Piazza site at http://piazza.com/jhu/spring2021/600649. 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 this course, you must be honest and truthful, abiding by the *Computer Science Academic Integrity Policy*:

Cheating is wrong. Cheating hurts our community by undermining academic integrity, creating mistrust, and fostering unfair competition. The university will punish cheaters with failure on an assignment, failure in a course, permanent transcript notation, suspension, and/or expulsion. Offenses may be reported to medical, law or other professional or graduate schools when a cheater applies.

Violations can include cheating on exams, plagiarism, reuse of assignments without permission, improper use of the Internet and electronic devices, unauthorized collaboration, alteration of graded assignments, forgery and falsification, lying, facilitating academic dishonesty, and unfair competition. Ignorance of these rules is not an excuse.

Academic honesty is required in all work you submit to be graded. Except where the instructor specifies group work, you must solve all homework and programming assignments without the help of others. For example, you must not look at anyone else's solutions (including program code) to your homework problems. However, you may discuss assignment specifications (not solutions) with others to be sure you understand what is required by the assignment.

If your instructor permits using fragments of source code from outside sources, such as your textbook or on-line resources, you must properly cite the source. Not citing it constitutes plagiarism. Similarly, your group projects must list everyone who participated.

Falsifying program output or results is prohibited.

Your instructor is free to override parts of this policy for particular assignments. To protect yourself: (1) Ask the instructor if you are not sure what is permissible. (2) Seek help from the instructor, TA or CAs, as you are always encouraged to do, rather than from other students. (3) Cite any questionable sources of help you may have received.

On every exam, you will sign the following pledge: "I agree to complete this exam without unauthorized assistance from any person, materials or device. [Signed and dated]". Your course instructors will let you know where to find copies of old exams, if they are available.

In addition, the specific ethics guidelines for this course are:

- 1. In the completion of individual homework assignments, you may not discuss your approach with or show specifics of your code to others. This includes fellow students, former students, friends, etc. You are permitted to request assistance from course staff (instructors, TAs and CAs) only.
- 2. You are permitted and expected to reuse and adapt code from lectures and the assigned texts in completing your projects. However, all original sources must be cited in comments within your code.
- 3. In using Piazza to ask questions about homework assignments, you should post privately to Instructors any questions that involve code or that would give away your approach to solving the assignment. Otherwise, you are encouraged to ask general, abstract questions, and post them publicly, so other students may benefit from from the discussion.

Report any violations you witness to the instructor.

You can find more information about university misconduct policies on the web at these sites:

- For undergraduates: <a href="https://e-catalogue.jhu.edu/arts-sciences/full-time-residential-programs/undergraduate-policies/student-life-policies/">https://e-catalogue.jhu.edu/arts-sciences/full-time-residential-programs/undergraduate-policies/student-life-policies/</a>
- For graduate students: <a href="https://e-catalogue.jhu.edu/arts-sciences/full-time-residential-programs/graduate-policies/student-life-policies/">https://e-catalogue.jhu.edu/arts-sciences/full-time-residential-programs/graduate-policies/</a>

### **Personal Wellbeing**

- Because of the ongoing COVID-19 pandemic special requirements may be in effect this term, and these may vary during the term. Please keep updated with these at the following sites:
  - o University information: <a href="https://covidinfo.jhu.edu/">https://covidinfo.jhu.edu/</a>
  - 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.
- The Johns Hopkins COVID-19 Call Center (JHCCC), which can be reached at 443-287-8500 seven days a week from 7 a.m. to 7 p.m., supports all JHU students, faculty, and staff experiencing COVID-19 symptoms.
   Primarily intended for those currently within driving distance of Baltimore, the JHCCC will evaluate your symptoms, order testing if needed,

- and conduct contact investigation for those affiliates who test positive. More information on the JHCCC and testing is on the <u>coronavirus</u> information website.
- 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 <a href="Student Health and Wellness">Student Health and Wellness</a>
  <a href="Center">Center</a> 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 <a href="https://studentaffairs.jhu.edu/student-life/student-outreach-support/absences-from-class/illness-note-policy/">https://studentaffairs.jhu.edu/student-life/student-outreach-support/absences-from-class/illness-note-policy/</a>
- All students with disabilities who require accommodations for this course should contact me at their earliest convenience to discuss their specific needs. If you have a documented disability, you must be registered with the JHU Office for Student Disability Services (101 Shaffer Hall; 410-516-4720; <a href="http://web.jhu.edu/disabilities/">http://web.jhu.edu/disabilities/</a>) to receive accommodations.
- Students who are struggling with anxiety, stress, depression or other mental health related concerns, please consider connecting with resources through the JHU Counseling Center. The Counseling Center will be providing services remotely to protect the health of students, staff, and communities. Please reach out to get connected and learn about service options based on where you are living this fall at 410-516-8278 and online at <a href="http://studentaffairs.jhu.edu/counselingcenter/">http://studentaffairs.jhu.edu/counselingcenter/</a>.
- 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: <a href="https://studentaffairs.jhu.edu/student-life/student-outreach-support/">https://studentaffairs.jhu.edu/student-life/student-outreach-support/</a> or email <a href="mailto:deanofstudents@jhu.edu">deanofstudents@jhu.edu</a>, 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**

As your instructor, 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 to seek mutually acceptable resolutions and accommodations. Reporting will never impact your course grade. You may also share concerns with Randal Burns, Department Head of Computer Science (randal@cs.jhu.edu) the Assistant Dean for Diversity and Inclusion (Darlene Saporu, dsaporu@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.

### **University Policy on Incompletes**

There are important revisions to the Incomplete Grade policy in effect for **UNDERGRADUATES** for the 2022-2023 academic year. 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 change here 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 now. The other significant change is the timeline for completion of an incomplete grade, now set at 45 calendar days after the last day of classes. Formerly,

the default deadline was the end of the third week 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**

Students may add a course up to **September 9, 2022** (independent academic work such as research may be added until **October 9, 2022**). They may drop courses up until **October 9, 2022** provided they remain registered for a minimum of 12 credits. Between **October 10, 2022** and **November 11, 2022** 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.

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