INFO B474
Next Generation Sequencing Data Analysis

Department of BioHealth Informatics
Indiana University School of Informatics and Computing, Indianapolis
Spring 2018

Section No.: Credit Hours: 3
Day/Time: Mondays, 3–5:40 pm
Location: WK 321, Walker Plaza Building
719 Indiana Avenue, Indianapolis, IN 46202 [map]
May have guest lectures, not necessarily in the same room and time
First Class: January 8, 2018
Website: Distribution of homework assignments and class notes via Canvas
Syllabus, outline of lectures, course calendar, and suggested reading will also be available from instructor’s website http://www.iupui.edu/~jangalab/
Final projects and presentation schedules

Instructor: Sarath Chandra Janga, Ph.D., Assistant Professor, Bioinformatics
Office Hours: Tuesdays and Thursdays, 11 am–12 pm or by Appointment
Office: WK 309, Walker Plaza Building
719 Indiana Avenue, Indianapolis, IN 46202 [map]
Phone: (317) 278-4147 (Office)
Email: jangalab@iupui.edu
Website: http://www.iupui.edu/~jangalab/

Prerequisites: INFO B473 or basic knowledge of programming, R, and Unix system management

COURSE DESCRIPTION
This course covers basic concepts of genomic sequencing datasets from several sequencing platforms, including how the data motivates computational needs and methods for analysis. Students learn how to devise approaches for analyzing massive clinical and biomedical sequencing datasets and for developing sound hypotheses and predictions from them.

EXTENDED COURSE DESCRIPTION
This course covers basic genomic sequencing analysis to give a foundation in sequencing technologies and their data analytics by introducing various existing implementations for specific tasks. This course covers concepts of a variety of genomic sequencing datasets
emerging in the post-genomic era from several sequencing platforms. It gives a background in molecular biology and informatics on how the generated data motivates computational needs and tasks for analysis. Upon completion, students will describe deft solutions to analyze a wide range of clinical and biomedical sequencing datasets to provide sound hypotheses and predictions from massive amounts of data.

**Prerequisites:** Knowledge of molecular biology and basic biochemistry is expected, although several of the required concepts will be taught throughout the course. Programming skills are expected and are a prerequisite for this course. Knowledge of programming fundamentals is essential as the instructor will be introducing a wide range of existing tools and approaches to analyze sequencing datasets and will be anticipating the students to either use or integrate the approaches to address specific questions in the assignments. To work with the datasets, knowledge of Unix-based system administration, Python or Perl programming, and R statistical analysis is expected. Knowledge of MySQL database management is helpful. If you are uncertain, course [INFO B473 Application Programming for Biomedical Data Analysis](#) is recommended before taking this course.

**Course structure:** The instructor introduces various types of sequencing datasets and applications commonly used for analyzing them in the first 10 weeks. Then each student presents a couple articles published within the last four years and a project on a particular theme or problem and submits a project report, which is also presented orally.

**Teaching and Learning Methods:** Class lectures and lab sessions

**Assessment methods:** Homework, midterm, project, presentation

**Required Readings:**

Readings and course notes are distributed via the course website.

**Recommended Readings:**

The following books are recommended for course assignments, exercises, and projects:


**Software used:**

Unix operating system, R, Python or Perl, and SQL (either Oracle and MySQL), Canvas, Microsoft Office
**Student Learning Outcomes:**

Upon completion of this course, students will

<table>
<thead>
<tr>
<th>RBT</th>
<th>PUL</th>
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<td>2</td>
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<td>A1</td>
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<td>3</td>
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<td>6</td>
<td>1C 2</td>
<td>F P</td>
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RBT: Revised Bloom’s Taxonomy; A: Assignment; M: Midterm; F: Final Project; P: Presentation

**Principles of Undergraduate Learning (PUL):**

Learning outcomes are assessed in the following areas:

1A. Core communication: written, oral and visual skills
1B. Core communication: quantitative skills
1C. Core communication: information resources skills
2. Critical thinking
3. Integration and application of knowledge
4. Intellectual depth, breadth, and adaptiveness
5. Understanding society and culture
6. Values and ethics

Major emphasis

Moderate emphasis
Assessments:

A1–6  *Programming assignments* (× 6)  60%
M1  *Midterm exam* (× 1)  20%
F  *Final Project* (× 1)  10%
P  *Presentation* (× 1)  10%

Grading Information:

- **Participation**: One point shall be deducted for each absence of the class from the final score. At most 5 points will be deducted for absences.
- **Homework/Assignment**: At least one assignment each on using types of sequencing datasets (available from canvas website). Strictly no copying of code from others and all the code should be functional to be awarded any grade.
- **Project**: Will comprise of a project for which you can work in groups of up to 2 students. Presentation of the project would be towards the end of the semester and will be evaluated as a group.

**Final project and presentation:**

Tackle a research problem, perform a literature review to identify relevant papers, present the papers, demonstrate the project, and write-up a short report; work individually or in pairs.

**Grading Scale:**

- A+ 97–100%  Professional level work, showing highest level of achievement, given only at the instructor’s discretion
- A 93–96.99%  Extraordinarily high achievement, quality of work; shows command of the subject matter
- A– 90–92.99%  Excellent and thorough knowledge of the subject matter
- B+ 87–89.99%  Above average understanding of material and quality of work
- B 83–86.99%  Mastery and fulfillment of all course requirements; good, acceptable work
- B– 80–82.99%  Satisfactory quality of work
- C+ 77–79.99%  Modestly acceptable performance and quality of work
- C 73–76.99%  Minimally acceptable performance and quality of work
- C– 70–72.99%  Unacceptable work (Core course must be repeated for credit)
- D+ 67–69.99%  Unacceptable work (Course must be repeated for credit)
- D 63–66.99%  Unacceptable work
- D– 60–62.99%  Unacceptable work
- F Below 60  Unacceptable work

No credit is granted for a grade below C.
<table>
<thead>
<tr>
<th>Week</th>
<th>Topics Covered</th>
<th>Assignments</th>
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<tbody>
<tr>
<td>1</td>
<td>Outline, syllabus, evaluation procedures and primer on molecular biology and sequencing platforms.</td>
<td>A1: Assemble a bacterial genome given short-read data.</td>
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<tr>
<td>2</td>
<td>Next generation DNA sequencing platforms, quality control, alignment (problems and solutions), Assembly of genomes (computational approaches)</td>
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<td>3</td>
<td>RNA-sequencing protocols, data storage and file formats, various aligners, expression quantification methods, differential expression analysis</td>
<td>A2: Given two GEO fastq datasets, identify and analyze the differentially expressed transcripts.</td>
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<td>4</td>
<td>Microbial DNA and RNA sequencing, computational approaches for assembly, quantification and detection of operons and transcription units. Introduction to Genome browsers.</td>
<td>A3: Given a fastq raw reads for a bacterial genome, align, quantify and construct the transcriptome map of the genome.</td>
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<td>5</td>
<td>Metagenomics and Metatranscriptomics: Computational methods to assemble, taxonomic mapping, analyze and study microbial abundance, interactions and pathways.</td>
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<td>6</td>
<td>ChIP-sequencing and related approaches. Computational approaches and analysis. Concept of peak calling and benchmarking studies illustrating the variations between methods.</td>
<td>A4: Analyze raw ChIP-seq data for a TF across cell types from encode data and analyze the differences.</td>
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<td>7</td>
<td>CLIP-sequencing and related approaches for building post-transcriptional networks. Computational approaches and analysis.</td>
<td>Midterm Exam</td>
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<td>8</td>
<td>Misc datasets: Small RNA sequencing/ protein occupancy profiling/ polyA sequencing</td>
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<td>9</td>
<td>Concept of genetic and somatic variation. Variant calling approaches. Issues and Considerations.</td>
<td>A5: Identify and delineate the germline and somatic SNPs from a series of samples taken from population sequencing project.</td>
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<td>10</td>
<td>Expression Quantitative Trait Loci identification/ Ethical considerations of personalized sequencing projects.</td>
<td>A6: Perform an eQTL analysis using a matched raw DNA and RNA sequencing dataset for a human tissue.</td>
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<td>11</td>
<td>The ENCODE consortium and major observations</td>
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<tr>
<td>12</td>
<td>Mining The Cancer Genome Atlas (TCGA) for personalized cancer medicine</td>
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EXPECTATIONS, GUIDELINES, AND POLICIES

Attendance:
A basic requirement of this course is that you will participate in all class meetings, whether online or face-to-face, and conscientiously complete all required course activities and assignments. Class attendance is required for classroom-based courses. It entails being present and attentive for the entire class period. Attendance shall be taken in every class. If you do not sign the attendance sheet while in class, you shall be marked absent. Signing the attendance sheet for another student is prohibited. The instructor is required to submit to the Registrar a record of student attendance, and action shall be taken if the record conveys a trend of absenteeism.

Only the following are acceptable excuses for absences: death in the immediate family (e.g. mother, father, spouse, child, or sibling), hospitalization or serious illness; jury duty; court ordered summons; religious holiday; university/school coordinated athletic or scholastic activities; an unanticipated event that would cause attendance to result in substantial hardship to one’s self or immediate family. Absences must be explained with the submission of appropriate documentation to the satisfaction of the instructor, who will decide whether missed work may be made up. Absences that do not satisfy the above criteria are considered unexcused. To protect your privacy, doctor’s excuses should exclude the nature of the condition and focus instead on how the condition impacts your attendance and academic performance.

Missing class reduces your grade through the following grade reduction policy: You are allowed two excused or unexcused absences. Each additional absence, unless excused, results in a 5% reduction in your final course grade. More than six absences result in an F in the course. Missing class may also reduce your grade by eliminating opportunities for class participation. For all absences, the student is responsible for all covered materials and assignments.

Incomplete:
The instructor may assign an Incomplete (I) grade only if at least 75% of the required coursework has been completed at passing quality and holding you to previously established time limits would result in unjust hardship to you. All unfinished work must be completed by the date set by the instructor. Left unchanged, an Incomplete automatically becomes an F after one year. http://registrar.iupui.edu/incomp.html

Deliverables:
You are responsible for completing each deliverable (e.g., assignment, quiz) by its deadline and submitting it by the specified method. Deadlines are outlined in the syllabus or in supplementary documents accessible through OnCourse. Should you miss a class, you are
still responsible for completing the deliverable and for finding out what was covered in class, including any new or modified deliverable. In fairness to the instructor and students who completed their work on time, a grade on a deliverable shall be reduced 10%, if it is submitted late and a further 10% for each 24-hour period it is submitted after the deadline.

CODE OF CONDUCT

All students should aspire to the highest standards of academic integrity. Using another student’s work on an assignment, cheating on a test, not quoting or citing references correctly, or any other form of dishonesty or plagiarism shall result in a grade of zero on the item and possibly an F in the course. Incidences of academic misconduct shall be referred to the Department Chair and repeated violations shall result in dismissal from the program.

All students are responsible for reading, understanding, and applying the Code of Student Rights, Responsibilities and Conduct and in particular the section on academic misconduct. Refer to The Code > Responsibilities > Academic Misconduct at http://www.indiana.edu/~code/. All students must also successfully complete the Indiana University Department of Education “How to Recognize Plagiarism” Tutorial and Test. https://www.indiana.edu/~istd You must document the difference between your writing and that of others. Use quotation marks in addition to a citation, page number, and reference whenever writing someone else’s words (e.g., following the Publication Manual of the American Psychological Association). To detect plagiarism instructors apply a range of methods, including Turnitin.com. http://www.ulib.iupui.edu/libinfo/turnitin

Academic Misconduct:

1. **Cheating:** Cheating is considered to be an attempt to use or provide unauthorized assistance, materials, information, or study aids in any form and in any academic exercise or environment.
   a. A student must not use external assistance on any “in-class” or “take-home” examination, unless the instructor specifically has authorized external assistance. This prohibition includes, but is not limited to, the use of tutors, books, notes, calculators, computers, and wireless communication devices.
   b. A student must not use another person as a substitute in the taking of an examination or quiz, nor allow other persons to conduct research or to prepare work, without advanced authorization from the instructor to whom the work is being submitted.
   c. A student must not use materials from a commercial term paper company, files of papers prepared by other persons, or submit documents found on the Internet.
   d. A student must not collaborate with other persons on a particular project and submit a copy of a written report that is represented explicitly or implicitly as the student’s individual work.
   e. A student must not use any unauthorized assistance in a laboratory, at a computer terminal, or on fieldwork.
   f. A student must not steal examinations or other course materials, including but not limited to, physical copies and photographic or electronic images.
g. A student must not submit substantial portions of the same academic work for credit or honors more than once without permission of the instructor or program to whom the work is being submitted.

h. A student must not, without authorization, alter a grade or score in any way, nor alter answers on a returned exam or assignment for credit.

2. Fabrication: A student must not falsify or invent any information or data in an academic exercise including, but not limited to, records or reports, laboratory results, and citation to the sources of information.

3. Plagiarism: Plagiarism is defined as presenting someone else’s work, including the work of other students, as one’s own. Any ideas or materials taken from another source for either written or oral use must be fully acknowledged, unless the information is common knowledge. What is considered “common knowledge” may differ from course to course.
   a. A student must not adopt or reproduce ideas, opinions, theories, formulas, graphics, or pictures of another person without acknowledgment.
   b. A student must give credit to the originality of others and acknowledge indebtedness whenever:
      1. directly quoting another person’s actual words, whether oral or written;
      2. using another person’s ideas, opinions, or theories;
      3. paraphrasing the words, ideas, opinions, or theories of others, whether oral or written;
      4. borrowing facts, statistics, or illustrative material; or
      5. offering materials assembled or collected by others in the form of projects or collections without acknowledgment

4. Interference: A student must not steal, change, destroy, or impede another student’s work, nor should the student unjustly attempt, through a bribe, a promise of favors or threats, to affect any student’s grade or the evaluation of academic performance. Impeding another student’s work includes, but is not limited to, the theft, defacement, or mutilation of resources so as to deprive others of the information they contain.

5. Violation of Course Rules: A student must not violate course rules established by a department, the course syllabus, verbal or written instructions, or the course materials that are rationally related to the content of the course or to the enhancement of the learning process in the course.

6. Facilitating Academic Dishonesty: A student must not intentionally or knowingly help or attempt to help another student to commit an act of academic misconduct, nor allow another student to use his or her work or resources to commit an act of misconduct.

OTHER POLICIES

1. Administrative withdrawal: Students must participate in all class discussions and conscientiously complete all required course activities and/or assignments. If a student is unable to attend, participate in, or complete an assignment on time, the student must inform the instructor. If a student misses more than half of the required activities within
the first 25% of the course without contacting the instructor, the student may be administratively withdrawn from this course. Administrative withdrawal may have academic, financial, and financial aid implications. Administrative withdrawal occurs after the full refund period, and a student who has been administratively withdrawn is ineligible for a tuition refund.

2. **Civility:** To maintain an effective and inclusive learning environment, it is important to be an attentive and respectful participant in lectures, discussions, group work, and other classroom exercises. Thus, unnecessary disruptions should be avoided, such as ringing cell phones, engagement in private conversations, and other unrelated activities. Cell phones, media players, or any noisy devices should be turned off during a class. Texting, web surfing, and posting to social media are generally not permitted. Laptop use may be permitted if it is used for taking notes or conducting class activities. Students should check with the instructor about permissible devices in class. IUPUI nurtures and promotes “a campus climate that seeks, values, and cultivates diversity in all of its forms and that provides conditions necessary for all campus community members to feel welcomed, supported, included, and valued” (IUPUI Strategic Initiative 9). IUPUI prohibits “discrimination against anyone for reasons of race, color, religion, national origin, sex, sexual orientation, marital status, age, disability, or veteran status” (Office of Equal Opportunity). Profanity or derogatory comments about the instructor, fellow students, invited speakers or other classroom visitors, or any members of the campus community shall not be tolerated. A violation of this rule shall result in a warning and, if the offense continues, possible disciplinary action.

3. **Communication:** For classroom-based courses, the instructor or teaching assistant should respond to emails by the end of the next class or, for online courses, within two Indiana University working days, which excludes weekends and holidays. The instructor should provide weekly office hours or accept appointments for face-to-face, telephone, or teleconferenced meetings, and announce periods of extended absence in advance.

4. **Counseling and Psychological Services (CAPS):** Students seeking counseling or other psychological services should contact the CAPS office at 274-2548 or capsindy@iupui.edu. For more information visit http://life.iupui.edu/caps/.

5. **Course evaluations:** Course evaluations provide vital information for improving the quality of courses and programs. Students are urged to complete one course and instructor evaluation for each section in which they are enrolled at the School of Informatics and Computing with the following exceptions: (a) The student has withdrawn from the course; (b) fewer than five students are enrolled in the section (in which case maintaining anonymity is difficult); and (c) the section is a laboratory that must be taken with a course having a different section number. Course evaluations are completed at https://soic.iupui.edu/app/course-eval/. Course evaluations are typically open from the eleventh week. Course evaluations are anonymous, which means that no one can view the name of the student completing the evaluation. In addition, no one can view the evaluation itself until after the instructor has submitted the final grades. In small sections, demographic information should be left blank, if it could be used to identify the student.

6. **Disabilities policy:** All qualified students enrolled in this course are entitled to
reasonable accommodations for a disability. Notify the instructor during the first week of class of accommodations needed. Students requiring accommodations register with Adaptive Educational Services (AES) and complete the appropriate AES-issued before receiving accommodations. The AES office is located at UC 100, Taylor Hall (Email: aes@iupui.edu, Tel. 317 274-3241). For more information visit http://aes.iupui.edu.

7. Email: Indiana University uses your IU email account as an official means of communication, and students should check it daily. Although you may have your IU email forwarded to an outside email account, please email faculty and staff from your IU email account.

8. Emergency preparedness: Know what to do in an emergency so that you can protect yourself and others. For more information, visit the emergency management website at http://protect.iu.edu/emergency.

9. IUPUI course policies: A number of campus policies governing IUPUI courses may be found at the following link: http://registrar.iupui.edu/course_policies.html

10. No class attendance without enrollment. Only those who are officially enrolled in this course may attend class unless enrolled as an auditor or making up an Incomplete by prior arrangement with the instructor. This policy does not apply to those assisting a student with a documented disability, serving in an instructional role, or administrative personnel. http://registrar.iupui.edu/official-enrollment-class-attendance.html Children may not attend class with their parents, guardians, or childcare providers.

11. Religious holidays: Students seeking accommodation for religious observances must submit a request form to the course instructor by the end of the second week of the semester. For information visit http://registrar.iupui.edu/religious.html.

12. Right to revise: The instructor reserves the right to make changes to this syllabus as necessary and, in such an event, will notify students of the changes immediately.

13. Sexual misconduct: IU does not tolerate sexual harassment or violence. For more information and resources, visit http://stopsexualviolence.iu.edu/.

14. Student advocate: The Student Advocate assists students with personal, financial, and academic issues. The Student Advocate is in the Campus Center, Suite 350, and may also be contacted at 317 274-4431 or studvoc@iupui.edu. For more information visit http://studentaffairs.iupui.edu/advocate.

MISSION STATEMENT

The Mission of IUPUI is to provide for its constituents excellence in

• Teaching and Learning;
• Research, Scholarship, and Creative Activity; and
• Civic Engagement.

With each of these core activities characterized by

• Collaboration within and across disciplines and with the community;
• A commitment to ensuring diversity; and
• Pursuit of best practices.
IUPUI’s mission is derived from and aligned with the principal components—Communities of Learning, Responsibilities of Excellence, Accountability and Best Practices—of Indiana University’s Strategic Directions Charter.

STATEMENT OF VALUES
IUPUI values the commitment of students to learning; of faculty to the highest standards of teaching, scholarship, and service; and of staff to the highest standards of service. IUPUI recognizes students as partners in learning. IUPUI values the opportunities afforded by its location in Indiana’s capital city and is committed to serving the needs of its community. Thus, IUPUI students, faculty, and staff are involved in the community, both to provide educational programs and patient care and to apply learning to community needs through service. As a leader in fostering collaborative relationships, IUPUI values collegiality, cooperation, creativity, innovation, and entrepreneurship as well as honesty, integrity, and support for open inquiry and dissemination of findings. IUPUI is committed to the personal and professional development of its students, faculty, and staff and to continuous improvement of its programs and services.