

University of Pittsburgh
Graduate School of Public Health
Department of Human Genetics
HUGEN 2010
Bioinformatic Resources for Geneticists

Spring 2023

Mondays · 03:00 PM–04:25 PM in 1501 Wesley W Posvar Hall

1 Credit

INSTRUCTOR

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COURSE DESCRIPTION

This course reviews the online bioinformatics resources available to geneticists. Students will learn to locate and use such resources and interpret the data therein to inform the development of research questions, aid in clinical decision-making, and enhance the understanding and contextualization of research results.

LEARNING OBJECTIVES

Upon completion of this course, the student will be able to:

- Name and locate online resources for querying databases of
 - life science and biomedical references and abstracts
 - NIH funded projects
 - phenotypes and genetic disorders
 - genetic variation
 - clinical assessments of genetic variation
 - ontologies
 - biological pathways and networks
 - comparative genomics
 - model organism variation and associated phenotypes
 - gene expression profiles
 - expression quantitative trait loci
 - regulatory elements evidence
 - sets of correlated variants
 - genotype–phenotype associations
 - tumor cell genetic variation
 - protein features
- Query each online resource for information that might be used in research question development or research and clinical finding interpretation.

COURSE PREREQUISITES / COREQUISITES

HUGEN 2022 · Human Population Genetics

HUGEN 2040 · Molecular Basis of Human Inherited Disease

LEARNING ACTIVITIES

Class sessions will consist of 40 minutes of lecture followed by 50 minutes of interactive lab. Lab time will be hands-on online activities scaffolded to advance from a cookbook activity with a well-defined answer to open-ended searches that simulate students' future graduate and professional activities and questions.

LEARNING MANAGEMENT SYSTEM

This course uses Canvas (canvas.pitt.edu) as its learning management system.

Accessibility

Ensuring an accessible and pleasant experience to all users, regardless of disability, is a key focus of Canvas. The Canvas platform was built using the most modern HTML and CSS technologies and is committed to W3C's Web Accessibility Initiative and [§508](#) guidelines.

COURSE MATERIALS**Required Equipment and Software**

All students will need access to a computer with a web browser (Chrome, Safari, Firefox, Internet Explorer, Opera) and the Internet. Students who do not have a computer may borrow a laptop from the Department of Human Genetics for use. Contact Dr. Fan for more information.

Reading

There is no required textbook for this course. Recommended and optional readings will be made available through Canvas.

ADDITIONAL RESOURCES

Additional bioinformatics resources, software and training can be found through the Health Sciences Library (www.hsls.pitt.edu); the Molecular Biology Information Service of the Health Science Library System (hsls.pitt.edu/molbio); and the school's dedicated librarian (Helena VonVille - <https://www.hsls.pitt.edu/staff/helena-vonville>)

You could also submit a [help ticket](#) to Molecular Biology Information Service via HSLs for additional information.

EVALUATION AND GRADING

All evaluations are graded pass/fail. Evaluation will be based on the following components:

Online Syllabus Review

There will be one (1) online syllabus review consisting of a series of questions posed online through a Google Form about the syllabus.

The online syllabus review is passed by correctly answering 80% of its tasks. If the online syllabus review is failed, it may be retaken as many times as necessary to pass it before the end of the University add/drop period (in Spring 2023, that is Fri Jan. 20th). Retaking the syllabus review does not use mulligans (described in the late policy below).

The initial deadline for submission of the online syllabus review is Mon Jan. 16th at 03:00 PM. A failed syllabus review can be retaken until Fri Jan 20th at 05:00 PM EST.

Online Assignments

There will be twelve (12) online assignments consisting of a series of questions posed online through a Google Form. For each week's module, students will be tasked to find information using that resource or resources covered in the module.

An online assignment is passed by correctly answering 80% of its tasks.

Searching databases requires attention to detail, the search results for **rs13** and **rs31** are very different and will not necessarily be obviously wrong to you. Answers that differ in orthography, such as **chr13 at 100 bp** vs **13:100** are acceptable, but ones that differ like **chr13** and **chr31** are not. Outside the classroom, if your supervisor asked for information about chr13 and you came to them with information about chr31, you'd have to go back and get the correct information, and we are following a similar model here. (Of course, much less leeway might be given in your ultimate profession to such typos.) There is no partial credit, even for obvious typos.

The deadline for submission of online assignments is Monday at 03:00 PM EST before the class. For example, the online assignment for the module discussed on Mon Jan. 9th is due on Mon Jan 16th at 03:00 PM EST.

A late assignment is failed unless you use a mulligan (described in the late policy below).

In-Class Group Activity

There will be one (1) final in-class group activity. Students will be divided into groups of four or five. Each group will be tasked with answering a series of questions, via a shared Google Doc, about an assigned phenotype and genes and genetic variants related to it using the resources

reviewed in class. For each question the group will also be asked to identify the resource used to answer the question.

The in-class group activity is passed by correctly answering 80% of its questions.

Late Policy & Revision Policy · Mulligans

Each student begins the term with five (5) “mulligans.” Each mulligan can be used for
 (a) a 24-hour extension on an online assignment or
 (b) the opportunity to retake an online assignment.

Additional 24-hour extensions on an online assignment can be taken using additional mulligans. For example, an assignment turned in 50 hours late would use three mulligans—one for the first extension up to 24 hours after the deadline, one for the second up to 48 hours after the deadline, and a third for the third up for 72 hours after the deadline.

An online assignment can only be retaken once. The retaken assignment is due one (1) week after the original assignment was due.

No mulligans are necessary to retake the syllabus review.

No mulligans can be used for the in-class group activity.

It is not necessary to ask for permission to use your mulligans. It is helpful to notify the instructor when you elect to take them to assist in tracking your progress in with the course.

If exigent circumstances arise that require extensions or exceptions beyond this policy, please contact Dr. Fan at your earliest opportunity.

Grading

The grade for the class is determined by the number of online assignments that are passed and, additionally for an **A**, whether the group you are assigned to passes the in-class group activity. All students must pass the syllabus review to receive a D or higher.

For example, if you are striving for a B, you need to pass the online syllabus review and 10 of the 12 online assignments and you do not need to participate in the in-class group activity.

	D	C	B	A
Online Syllabus Review	1/1	1/1	1/1	1/1
Online Assignments	8/12	9/12	10/12	11/12
In-Class Group Activity				1/1

An **A+** can be earned by passing the online syllabus review, 11/12 online assignments, the in-class group activity, and by finishing the term with at least four unused mulligans.

SCHEDULE

Module	Date	Topic	Databases	Instructor
1	<i>Mon Jan 9</i>	Introduction & Literature Informatics	HSLs, NCBI, PubMed, MyNCBI, NIH RePORTER	Frank Fan
	<i>Mon Jan 16</i>	<i>No Class – Martin Luther King Day</i>		
2	<i>Mon Jan 23</i>	Genome Browsers 1	UCSC Genome Browser	Frank Fan
3	<i>Mon Jan 30</i>	Genome Browsers 2	UCSC Genome Browser	Frank Fan
4	<i>Mon Feb 6</i>	Variation Databases	dbSNP, gnomAD, BRAVO, SNPedia	Ryan Minster
5	<i>Mon Feb 13</i>	Phenotype–Genotype Associations	GWAS Catalog, PheGenI, genebase	Frank Fan
6	<i>Mon Feb 20</i>	Comparative Genomics, Data Integrators, & Accessibility	UCSC Genome Browser, MGI, VEP, Color Oracle	Ryan Minster
7	<i>Mon Feb 27</i>	Gene Regulation Polygenic Scores	GTEx, RegulomeDB, HaploReg PGS Catalog	Frank Fan
	<i>Mon Mar 6</i>	<i>No Class – Spring Break</i>		
8	<i>Mon Mar 13</i>	Cancer Genetics & Genomics	GDC (TCGA), COSMIC	Frank Fan
9	<i>Mon Mar 20</i>	Ontologies, Pathways & Networks	Gene Ontology, Pathway Commons, PANTHER, KEGG	Frank Fan
10	<i>*Mon Mar 27</i>	Clinical Relevance 1	ClinVar, OMIM	Michelle Morrow
11	<i>*Mon Apr 3</i>	Clinical Relevance 2	GTR, GeneTests	Michelle Morrow
12	<i>Mon Apr 10</i>	Proteomics Miscellany	UniProt, String, alphafold	Frank Fan
	<i>Mon Apr 17</i>	In-Class Group Activity		
	<i>Mon Apr 24</i>	<i>No Class</i>		

* Dates might be changed due to the guest speaker's availability.

ACADEMIC POLICIES

Academic Integrity

All students are expected to adhere to the school's standards of academic honesty. Cheating/plagiarism will not be tolerated. The Graduate School of Public Health's policy on academic integrity, which is based on the University policy, is available online in the Pitt Public Health Academic Handbook www.publichealth.pitt.edu/home/academics/academic-requirements. The policy includes obligations for faculty and students, procedures for adjudicating violations, and other critical information. Please take the time to read this policy.

Plagiarism

University policy:

Integrity of the academic process requires that credit be given where credit is due. Accordingly, it is unethical to present as one's own work the ideas, representations, words of another, or to permit another to present one's own work without customary and proper acknowledgement of sources.

A student has an obligation to exhibit honesty and to respect the ethical standards of the profession in carrying out his or her academic assignments. Without limiting the application of this principle, a student may be found to have violated this obligation if he or she:

- Presents as one's own, for academic evaluation, the ideas, representations, or words of another person or persons without customary and proper acknowledgment of sources.
- Submits the work of another person in a manner which represents the work to be one's own.

Source

To avoid plagiarism, you must give "customary and proper acknowledgment of sources" by appropriately and clearly identifying which thoughts are yours and which are others, and appropriately citing your sources.

Sophisticated plagiarism detection software will be used in this course. If plagiarism is detected, you will automatically receive a grade of zero for that assignment and the incident will be reported, as required, to your Dean.

Copyright Notice

These materials may be protected by copyright. United States copyright law, 17 USC section 101, et seq., in addition to University policy and procedures, prohibit unauthorized duplication or retransmission of course materials. See [Library of Congress Copyright Office](#) and the [University Copyright Policy](#).

Course Recording

This class or portions of this class will be recorded by the instructors for educational purposes. These recordings will be shared only with students enrolled in the course via Canvas and will be deleted at the end of the course.

To ensure the free and open discussion of ideas, students may not record classroom lectures, discussion and/or activities without the advance written permission of the instructor, and any such recording properly approved in advance can be used solely for the student's own private use.

Disability Resources

If you have a disability for which you are or may be requesting an accommodation, you are encouraged to contact both your instructor and [Disability Resources and Services](#) (DRS), 140 William Pitt Union, (412) 648-7890, drsrecep@pitt.edu, (412) 228-5347 for P3 ASL users, as early as possible in the term. DRS will verify your disability and determine reasonable accommodations for this course.

Sexual Misconduct, Required Reporting, & Title IX

The University is committed to combatting sexual misconduct. As a result, you should know that University faculty and staff members are required to report any instances of sexual misconduct, including harassment and sexual violence, to the University's Title IX office so that the victim may be provided appropriate resources and support options. What this means is that as your professor, I am required to report any incidents of sexual misconduct that are directly reported to me, or of which I am somehow made aware.

There are two important exceptions to this requirement about which you should be aware:

A list of the designated University employees who, as counselors and medical professionals, do not have this reporting responsibility and can maintain confidentiality, can be found here: <https://www.diversity.pitt.edu/civil-rights-title-ix/make-report/report-form>

An important exception to the reporting requirement exists for academic work. Disclosures about sexual misconduct that are shared as part of an academic project, classroom discussion, or course assignment, are not required to be disclosed to the University's Title IX office.

If you are the victim of sexual misconduct, Pitt encourages you to reach out to these resources:

- Title IX Office: 412-648-7860
- SHARE @ the University Counseling Center: 412-648-7930 (8:30 A.M. TO 5 P.M. M-F) and 412-648-7856 (AFTER BUSINESS HOURS)

If you have a safety concern, please contact the University of Pittsburgh Police, 412-624-2121.

Other reporting information is available here: <https://www.diversity.pitt.edu/civil-rights-title-ix-compliance/make-report>

Equity, Diversity, and Inclusion

The University of Pittsburgh Graduate School of Public Health considers the diversity of its students, faculty, and staff to be a strength and critical to its educational mission. Pitt Public Health is committed to creating and fostering inclusive learning environments that value human dignity and equity and promote social justice. Every member of our community is expected to be respectful of the individual perspectives, experiences, behaviors, worldviews, and backgrounds of others. While intellectual disagreement may be constructive, no derogatory statements, or demeaning or discriminatory behavior will be permitted.

If you feel uncomfortable or would like to discuss a situation, please contact any of the following: the course director or course instructor;

- the Pitt Public Health Associate Dean responsible for diversity and inclusion;
- the University's Office of Diversity and Inclusion at 412-648-7860 or
- <https://www.diversity.pitt.edu/civil-rights-title-ix/make-report/report-form>
(anonymous reporting form)