

HUGEN 2051: Inborn Errors of Development, Spring 2019

General information

Meeting time: Mondays and Thursdays, 5:00-6:00 PM
Location: 3140 Public Health
Credit Hours: 2

Instructors

Name	Office	email	phone	Office hours
Zsolt Urban, PhD	3130 PUBHL	urbanz@pitt.edu	412.648.8269	By appointment
Beth Roman, PhD	3132 PUBHL	romanb@pitt.edu	412.624.7006	By appointment

CourseWeb access

Login through the Pitt portal, <http://my.pitt.edu>, with your standard Pitt username and password.

If you are already logged into my.pitt.edu, you can access directly at <http://courseweb.pitt.edu>

On the right hand side, under "My Courses", click on the course title, [2194_HUGEN_2051_SEC1010_INBORN ERRORS OF DEVELOPMENT](#).

Catalogue Description

This course focuses on the connections between human development and inherited disease. The course will include core principles of development of the body plan and signaling pathways involved in development and differentiation. These biological processes will be used to categorize inherited human diseases, understand disease mechanisms, and the current efforts to develop targeted treatments.

Course rationale

Recent discoveries in developmental biology and human genetics have been rapid and synergistic. The use of animal models of development identified genes that were subsequently found to be causing congenital malformations in human individuals. Conversely, discovery of human disease genes using the tools developed by the human genome project and the understanding of genotype-phenotype correlations uncovered key molecular pathways of development. Traditionally, human genetics and developmental biology are taught as separate courses. This course is intended to explore the interdisciplinary overlap between these subjects.

Learning Objectives

By the end of this course, each student will be able to:

- Understand the development of major organ systems
- Apply knowledge of molecular signaling pathways directing human development
- Identify and classify inherited developmental diseases using molecular information
- Search the literature based on citations and impact factor
- Organize and summarize research data in writing and speech

Prerequisites

No prerequisites are required, but students will find the course easier if they had some prior undergraduate or graduate coursework in Developmental Biology or Molecular Biology. Taking HUGEN 2040 (Molecular Basis of Human Inherited Disease) prior to this course is recommended. For English as a Second Language students, a general academic writing course is also recommended.

Teaching Philosophy

This course is intended for upper level graduate students and places an emphasis on active participation. The classes will consist of lectures and article discussions. The assignments are designed

HUGEN 2051: Inborn Errors of Development, Spring 2019

to improve your scientific writing and presentation skills. We value your ability to think independently and critically and to see connections between seemingly disparate research findings.

Textbooks/Articles/Readings

- Gilbert SF, Barresi MJF (2016) *Developmental Biology* 11th Ed. ISBN 978-1-60535-470-5 *Although not required for the course, we recommend this text, as it provides an excellent overview of the topic. You can purchase this text via online retailers such as Amazon for a relatively low price. Earlier editions (9th, 10th) of this text are also fine.*
- Erickson RP, Wynshaw-Boris AJ (2016) *Epstein's Inborn Errors of Development*. 3rd Ed, ISBN-978-0-19-993452-2
This is a reference book for background reading on specific developmental disorders.

CourseWeb Organization

All readings and course material will be found on the CourseWeb site for this class. All information can be accessed via the course menu on the left-hand side of the home page

Course Documents houses all PowerPoint presentations, handouts, supplemental readings, homework, answer keys, etc. Each module has its own subfolder within the *Course Documents* folder.

Printed handouts will not be available in class. Although you may wish to use a laptop to view the PowerPoint presentation during class, we strongly encourage you to take hand-written notes.

Ground Rules for Class

Please be on time and turn off your cell phone. If you choose to use a laptop during class, make sure that the sound is off. Please stay focused: no texting or online chatting, game playing, or browsing during class.

Student Performance Evaluation (Assessments and Weights)

Written article commentary (50 points): You will need to select a list of 5 articles related to development and human disease. The articles need to be recent (not older than 1 year), must not have been published by University of Pittsburgh researchers, and each must be published in a journal with an impact factor of 6 or higher. For each article, list all review articles that cited it. This list is due on the 5th class (January 18) and counts for 10 points of the assignment. The subject of the commentary will be selected from this list of 5 articles (one of the 5 articles). The commentary should be in the "Perspectives" format of *Science* magazine (900-1100 words, 5-15 references) and should provide a context to the article within a scientific field and discuss its interdisciplinary significance. Rather than a simple summary of the article, the commentary should give a new dimension to the research study. Also point out the limitations of the study. An article that has been the main focus of prior reviews and commentaries may not be used for this assignment. The commentary is due on February 13. Grading guide: article list (10 points), accuracy of summary (10 points), discussion of context and significance (10 points), new dimension to research (10 points), accuracy and relevance of references (10 points). Late assignments will be docked at -2 points per day.

Mini-review (100 points): Select a recent, well-focused topic in the area of developmental genetics and write a 2000-3000 word mini review about the topic with 20-50 references. Choose a topic that you find exciting, important or particularly interesting. In the introduction, discuss previous reviews of the topic and state how your review is new or different from the previous ones. The mini-review is due on March 18. Grading guide: impact and importance of the topic (15 points), discussion of prior reviews and novelty of this review (10 points), logical structure (15 points), clear, engaging style (15 points), correct grammar and language (15 points), accuracy and relevance of references (15 points),

HUGEN 2051: Inborn Errors of Development, Spring 2019

discussion of future research directions and opportunities (15 points). Late assignments will be docked at -4 points per day.

Exam (100 points): The exam will consist of short questions focusing on the main ideas covered in the course and research problems/scenarios. The exam will be open notes.

Presentation (50 points): Two students will jointly prepare and present 30-minute powerpoint presentation of a recent high impact publication related to one of the lecture topics listed in the class schedule. The topic should be selected at the end of the first lecture and the publication by the 4th class (January 16). The publication should not be the same as the subject of your commentary or a subject of a previous Human Genetics Journal Club meeting. Your presentation should briefly introduce the research field and the question addressed by the publication. Summarize the results of the study and describe the conclusions. Your final few slides should include a criticism of the paper (Do the results support the conclusions? Are the methods appropriate? Are there any limitations?) As a general guideline, count 1 minute per slide. Images used in the presentation should be properly referenced. Grading guide: clarity and accuracy of the presentation (10 points), quality of visuals (10 points), critical evaluation (10 points), on-time delivery (10 points).

	Points	Percent of grade
Perspectives	50	17
Mini-review	100	33
Exam	100	33
Presentation	50	17
Total for course	300	100

Grading Scale

If the class mean for all completed work at the end of the semester is below 85% (B), the mean will be linearly adjusted to 85% and individual grades recalculated accordingly. *Grades on individual exams or assignments will not be adjusted.* The grading scale (below) will then be used to determine final grades.

A+	97-100%	A	93-96.99%	A-	90-92.99%
B+	87-89.99%	B	83-86.99%	B-	80-82.99%
C+	77-79.99%	C	73-76.99%	C-	70-72.99%
D+	67-69.99%	D	63-76.99%	D-	60-62.99%
F	59.99% and below				

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.

Disability Services

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,

HUGEN 2051: Inborn Errors of Development, Spring 2019

(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.

Accessibility

CourseWeb/Blackboard is ADA Compliant and has fully implemented the final accessibility standards for electronic and information technology covered by Section 508 of the Rehabilitation Act Amendments of 1998. Please note that, due to the flexibility provided in this product, it is possible for some material to inadvertently fall outside of these guidelines.

Sexual Misconduct, Required Reporting and Title IX Statement

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:

www.titleix.pitt.edu/report/confidentiality

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: www.titleix.pitt.edu/report-0

Diversity Statement

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. 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 instructor;
- the Pitt Public Health Associate Dean for Diversity at 412-624-3506 or nam137@pitt.edu;
- the University's Office of Diversity and Inclusion at 412-648-7860 or <https://www.diversity.pitt.edu/make-report/report-form> (anonymous reporting form).

Copyright Notice

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HUGEN 2051: Inborn Errors of Development, Spring 2019

or retransmission of course materials. See [Library of Congress Copyright Office](#) and the [University Copyright Policy](#).

Statement on Classroom Recording

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.

Library and Writing Center Services

As a University of Pittsburgh student, a wealth of resources for researching your project is available through the Health Sciences Library (www.hsls.pitt.edu) and the school's dedicated librarian (Barb Folb - <https://www.hsls.pitt.edu/staff/barbara-folb>). In addition, writing assistance is available through the University Writing Center (www.writingcenter.pitt.edu/).

HUGEN 2051: Inborn Errors of Development, Spring 2019

Date	Lecture	Topic	Assignment	Lecturer
1/07/19	1	Introduction		Urban
1/09/19	2	Fertilization		Urban
1/14/19	3	Early development and implantation		Urban
1/16/19	4	Axis determination and HOX genes	Commentary article list due Article selection for short presentation due	Urban
1/21/19		<i>No class-MLK Day</i>		
1/23/19	5	Nervous system		Urban
1/28/19	6	Neural crest		Urban
1/30/19	7	Eye		TBA
2/04/19	8	Ear		Urban
2/06/19	9	Somite and muscle		Urban
2/11/19	10	Cartilage and bone		Urban
2/13/19	11	Limbs	Perspectives due	Urban
2/18/19	12	Heart		Roman
2/20/19	13	Blood vessels		Roman
2/25/19	14	Kidney		TBA
2/27/19	15	Endodermal derivatives		Urban
3/04/19	16	Skin and appendages		Urban
3/06/19	17	Sex determination, gametes		Urban
3/11/19		<i>No class-Spring Break</i>		
3/13/19		<i>No class-Spring Break</i>		
3/18/19		Aging	Mini-review due	TBA
3/20/19		Stem cells		Roman
3/25/19		Environmental disruptors		Roman
3/27/19		Exam		
4/01/19		Article 1		
4/03/19		Article 2		
4/08/19		Article 3		
4/10/19		Article 4		
4/15/19		Article 5		
4/22/19		Article 6		