Human Molecular Genetics
BI 423/523

Lecture Time: 10:00-11:20 Monday & Wednesday
Lecture Location: 254 STB
Instructor: Jana Prikryl, jprikryl@uoregon.edu
Office Hours: Fridays at 10:00-11:00 am, KLA 65C

This 4 credit course consists of two 1:20 long lectures per week and counts towards:
- The BI420-499 BI/MARB major requirement
- The Human Biology Emphasis area
- The Molecular, Cellular, Developmental Biology Emphasis area
- The upper-division BI emphasis area for GS majors (if taken for a letter grade)

All materials and readings for this class are available on the course Canvas site.

Course description

We will work together to investigate current knowledge and technologies in human molecular genetics, including their medical applications and ethical considerations. This field is immense and continually growing; we only have time to scratch the surface during the 10 weeks we have together so a primary goal of this course is to provide you with the background, confidence, and tools you need to continue to investigate the areas of genetics that you are most interested in. This course relies heavily on group work and student presentation, and it will be critically important for students to come to class having completed the assigned reading/listening/viewing before class. **If you know you will need to miss a lecture please let me know during week 1.** This course relies on current review articles, videos, and audio files to introduce content, and primary articles to take a more in-depth look at several example concepts. All these materials are posted on the course Canvas site. Please bring your laptop/tablet (internet-enabled device) to lectures.

Learning Outcomes

Translational skills:
This course is structured to encourage you to develop skills in:
- Finding and evaluating scientific information
- Working productively as part of a team
- Identifying and communicating the main points of complex information in written and verbal forms
- Critical thinking
- Ethical reflection on current issues related to our continually increasing understanding of genetics and genetic manipulation.

Content knowledge:
This course will focus on the below content areas
- High-throughput sequencing: methodologies and applications
- Genome editing and the CRISPR Cas9 system
- Higher-order chromatin structure and how it is established
- Non-coding RNA and X-inactivation
- Manipulation of RNA processing for medical applications

Student workload

The bulk of the grade for this course will be based on 4 group projects that include an individual written component and a group presentation component. These projects will require you to investigate, in detail, one aspect of a much broader
genetics topic. You can expect to spend approximately 10 hours per project on doing research and preparing materials for the presentations (20 hours for graduate students). You can expect to spend approximately 5 hours a week outside of class on reading assigned articles, watching videos and listening to audio files. This course will rely heavily on group work and group presentation of information.

How grades will be determined

Each project will be worth 18 points (72pt total for all 4) as distributed below.

<table>
<thead>
<tr>
<th>Grade distribution</th>
<th>18pt total each</th>
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<tbody>
<tr>
<td>Group-work day:</td>
<td>2pt total each</td>
</tr>
<tr>
<td>Initial group work and presentation (group):</td>
<td>3pt</td>
</tr>
<tr>
<td>Intervening time:</td>
<td>8pt</td>
</tr>
<tr>
<td>Handout/Vocab: 8pt</td>
<td></td>
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<tr>
<td>Infographic (individual):</td>
<td>3pt</td>
</tr>
<tr>
<td>Additional sources (individual):</td>
<td>1pt</td>
</tr>
<tr>
<td>Vocab list (group):</td>
<td>1pt</td>
</tr>
<tr>
<td>Questions (group):</td>
<td>3pt</td>
</tr>
<tr>
<td>Presentation day:</td>
<td>4pt</td>
</tr>
<tr>
<td>Explanation of section &amp; timing:</td>
<td></td>
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<tr>
<td>Group self-assessment (individual):</td>
<td>1pt</td>
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The remaining 28 points will be based on assessments used to ensure that everyone comes to class having done the assigned preparatory work. These will take the form of short writing assignments or quizzes at the start of class.

Course policies

Accessible Education – (see https://aec.uoregon.edu/best-practices-faculty for more information)
The University of Oregon is working to create inclusive learning environments. Please notify me if there are aspects of the instruction or design of this course that result in disability-related barriers to your participation. You are also encouraged to contact the Accessible Education Center in 360 Oregon Hall at 541-346-1155 or uoaec@uoregon.edu.

Academic Misconduct – (See https://dos.uoregon.edu/academic-misconduct for more information)
The University Student Conduct Code (available at conduct.uoregon.edu) defines academic misconduct. Students are prohibited from committing or attempting to commit any act that constitutes academic misconduct. By way of example, students should not give or receive (or attempt to give or receive) unauthorized help on assignments or examinations without express permission from the instructor. Students should properly acknowledge and document all sources of information (e.g. quotations, paraphrases, ideas) and use only the sources and resources authorized by the instructor. If there is any question about whether an act constitutes academic misconduct, it is the students’ obligation to clarify the question with the instructor before committing or attempting to commit the act. Additional information about a common form of academic misconduct, plagiarism, is available at https://researchguides.uoregon.edu/citing-plagiarism.

Inclement Weather – It is generally expected that class will meet unless the University is officially closed for inclement weather. If it becomes necessary to cancel class while the University remains open, this will be announced on Canvas and by email. Updates on inclement weather and closure are also communicated in other ways described here: https://hr.uoregon.edu/about-hr/campus-notifications/inclement-weather/inclement-weather-immediate-updates

I am a student-directed employee. For information about my reporting obligations as an employee, please see Employee Reporting Obligations on the Office of Investigations and Civil Rights Compliance (OICRC) website. Students experiencing any form of prohibited discrimination or harassment, including sex or gender-based violence, may seek information and resources at safe.uoregon.edu, respect.uoregon.edu, or investigations.uoregon.edu or contact the non-confidential Title IX office/Office of Civil Rights Compliance (541-346-3123), or Dean of Students offices (541-346-3216), or call the 24-7 hotline 541-346-SAFE for help. I am also a mandatory reporter of child abuse. Please find more information at Mandatory Reporting of Child Abuse and Neglect.”
### Course schedule and assignments (subject to change)

<table>
<thead>
<tr>
<th>Mon January</th>
<th>Tue</th>
<th>Wed</th>
<th>Thu</th>
<th>Fri</th>
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</table>
| 6 **Intro to class**  
Individual introductions  
Intro the first paper and paper projects  
Annie, talk about library research | 7 | 8 **High throughput sequencing (HTS)**  
*Watch:* Sequencing video from BI 320 (unless you remember it)  
*Listen:* 2019 NPR story: HTS in medical diagnostics  
*Read:* 2015 HTS review, Molecular Cell (presentation paper) | 9 | 10 |
| **13 HTS presentations** | 14 | 15 **Human Genome**  
*Watch:* Genome organization video from BI 320 (unless you remember it)  
*Listen:* 2019 NPR story: genetic surveillance  
*Read:* 2019 Genomic surveillance, Nature  
*Read:* 2016 Genomic diversity, Nature  
*Read:* 2014 Functional DNA, PNAS | 16 | 17 |
| **20 MLK** | 21 | 22 **CRISPR Cas9 Gene editing**  
*Watch:* CRISPR Cas9 video from BI 320 (unless you remember it)  
*Read:* web: How Does Genome Editing Work?  
*Read:* 2018 CRISPR Cas9, Nature Communications (Presentation paper) | 23 | 24 |
| **27 Prime editing**  
*Read:* 2019 Prime Editing, Science  
*Read:* 2019 Prime Editing, Nature | 28 | 29 **Gene editing and ethics**  
*Watch:* John Oliver (skip the mouse/tick part)  
*Watch:* Video on Zolgensma site  
*Listen:* 2018 NPR story: gene-edited babies  
*Read:* 2018 Gene Therapy, Science  
Annie on finding reputable papers | 30 | 31 |
| Mon February | Tue | Wed | Thu | Fri |
| 3 **Editing presentations** | 4 | 5 **Chromatin Structure, Topologically Associating Domains (TADs)**  
*Read:* 2019 Genome folding into TADs, Science Advances (Presentation paper) | 6 | 7 |
| **10 Structure and disease**  
*Read:* 2019, Crom Topology in devo and disease, Curr Opin Gen and Dev | 11 | 12 **TADs Loop Extrusion**  
*Read:* 2018, Imaging loop extrusion, Science | 13 | 14 |
| **17 Structure presentations** | 18 | 19 **non-coding RNA (ncRNA)**  
*Read:* 2014 Rise of Regulatory RNAs, Nat Rev Gen (Presentation paper) | 20 | 21 |
| **24 X inactivation**  
*Read:* 2011 Reg of X inactivation, Nat Rev Gen | 25 | 26 **X inactivation**  
*Read:* 2015 X inactivation, Nature | 27 | 28 |
| Mon March | Tue | Wed | Thu | Fri |
| 2 **ncRNA presentations** | 3 | 4 **Splicing**  
*Read:* 2017 Targeting splicing in human disease, Genes | 5 | 6 |
| **9 Spinraza**  
*Read:* 2017 Spinraza, Nat Neuro  
*Read:* 2017 Spinraza, Transl Neuro | 10 | 11 **Bias and pain**  
*Watch:* John Oliver Clip  
*Watch:* retort to Oliver Clip  
*Read:* 2016 Bias and pain, PNAS | 12 | 13 |