

# Human Molecular Genetics

## BI 423/523

**Lecture Time:** 12:15-1:45 Tuesday & Thursday **on zoom** (access via canvas, password, if needed: BI423)

**Lecture Location:** Remote synchronous

**Instructor:** Jana Prikryl, [jprikryl@uoregon.edu](mailto:jprikryl@uoregon.edu)

**Office Hours:** 1:45 to 2:15 Tuesday & Thursday (or by appointment)

**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)

### Most important logistics things to know:

1. All materials and readings for this class are available on the course Canvas site.
2. Use the **Modules** section of Canvas to find out what is due and to access all readings/videos/audio files.
3. Best way to contact Jana is via regular old email: [jprikryl@uoregon.edu](mailto:jprikryl@uoregon.edu)
4. Class announcements will be made through the Canvas announcement feature.
5. Class will take place over zoom, please have cameras on if possible. If it's not possible please let me know.
6. If the internet goes down (on your end): reconnect when you can, if it's down for a long time let me know what happened once you are back up and running.
7. If the internet goes down (on my end):

try to re-enter the meeting right away and again in 1 min. In case I accidentally ended the meeting.

If that does not work check for an announcement from me within 10 min. In case the issue is limited to zoom and I can still use other parts of the internet.

If there are no announcements, meaning my connection is completely down, contact your group and discuss whatever we were talking about with them. I will eventually send an announcement (once I'm back up and running).

### 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.

## 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, verbal, and digital 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, mRNA vaccines

## 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 video 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 (which can be done at 2X speed, I recommend this for a lot of them) and listening to audio files. This course will rely heavily on group work and presentation of information. When surveyed at the end of the term, last year's class approximated the workload for this course to be either equivalent or even a little less than other 400 level BI courses.

## How grades will be determined

**This course has no final exam, or any other exam, aside from the low point reading assessments discussed below.**

**64 points:** 4 multimedia presentation projects (16 points each): Distribution and expectations are described in the Presentation Expectations document.

**36 points:** The remaining 36 points will be based on assessments (2-3 points each) 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 each class (except for first day of class and main presentation workdays (in calendar below).

## 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](mailto: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](https://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>.

*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](https://safe.uoregon.edu), [respect.uoregon.edu](https://respect.uoregon.edu), or [investigations.uoregon.edu](https://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) listed in recommended order**

Tues <b>January</b>	Thurs.
<b>5 Intro and info</b> Individual introductions Class introduction Evaluating information (popular and scientific) Read (after class): 2017 Wineburg McGrew	<b>7 High throughput sequencing (HTS)</b> <b>Watch:</b> 23 and me video OPB <b>Listen:</b> 2019 NPR story: HTS in medical diagnostics <b>Read:</b> 2016 Genomics diversity, Nature <b>Watch:</b> Sequencing video from BI 320 <b>Read:</b> 2015 HTS review, Molecular Cell (presentation paper)
<b>12</b> <b>Listen:</b> 2019 NPR story: genetic surveillance <b>Read:</b> 2019 Genomic surveillance, Nature <b>Read:</b> 2008 Gen Res in Native comm <b>Read:</b> 2017 Navajo gen res ban, Nature	<b>14 Human Genome</b> <b>Watch:</b> Genome organization video from BI 320 <b>Read:</b> 2014 Functional DNA, PNAS (Optional Supplement is also available)
<b>19 HTS presentations workday no reading assessment today</b>	<b>21 CRISPR Cas9 Gene editing</b> <b>Listen:</b> NPR short story on Editing <b>Watch:</b> CRISPR Cas9 video from BI 320 <b>Watch:</b> Daudna talk <b>Read:</b> web: How Does Genome Editing Work? <b>Read:</b> 2018 CRISPR Cas9, Nature Communications (Presentation paper)
<b>26 Prime editing</b> <b>Watch:</b> Gene Drive short video <b>Optional Read:</b> 2019 CRISPR detection tools, Nature <b>Read:</b> 2019 Prime Editing, Science <b>Read:</b> 2019 Prime Editing, Nature	<b>28 Gene editing and ethics</b> <b>Watch:</b> John Oliver (skip the mouse/tick part) <b>Watch:</b> Video on Zolgensma site <b>Listen:</b> 2018 NPR story: gene-edited babies <b>Read:</b> 2019 BBC update on gene-edited babies <b>Read:</b> 2017 Gene Therapy Threat, Impact Ethics <b>Read:</b> 2018 Gene Therapy, Science

Tues <b>February</b>	Thurs.
<b>2 Editing presentations workday no reading assessment today</b>	<b>4 Chromatin Structure, Topologically Associating Domains (TADs)</b> <b>Read:</b> 2019 Genome folding into TADs, Science Advances (Presentation paper)
<b>9 Structure and disease</b> <b>Read:</b> 2019, Crom Topology in devo and disease, Curr Opin Gen and Dev	<b>11 TADs Loop Extrusion</b> <b>Read:</b> 2018, Imaging loop extrusion, Science
I'm still working out the details of the order of stuff below this point. All the papers are there, just not sure when we will read each one.	
<b>16 Structure presentations workday no reading assessment today</b>	<b>18 non-coding RNA (ncRNA) OR SARS CoV 2</b> <b>Read:</b> 2014 Rise of Regulatory RNAs, Nat Rev Gen (Presentation paper)
<b>23 SARS CoV 2- identification</b> <b>Watch:</b> John Oliver video on Conspiracy theories <b>Read:</b> 2020 SARS nCoV characterization, The Lancet (optional supplement also available)	<b>25 SARS CoV 2- Diagnostics, or symptoms (not sure yet)</b> <b>Read:</b> 2020 CoV2 testing, Biosensors Bioelectronics

Tues <b>March</b>	Thurs.
<p>2 ncRNA <b>presentations workday no reading assessment today</b></p>	<p>4 <b>SARS CoV 2-Vaccination</b>          There is a John Oliver video on vaccines that you can look up if you want, not mandatory.  <b>Read:</b> 2020 Ethics Vaccine, Science  <b>Read:</b> 2020 nCoV 2 Vaccines, Nature</p>
<p>9 <b>Splicing Spinraza</b>  <b>Read:</b> 2017 Spinraza, Nat Neuro  <b>Read:</b> 2017 Spinraza, Transl Neuro  <b>Read:</b> 2017 Targeting splicing in human disease, Genes</p>	<p>11 <b>Bias and pain</b>  <b>Watch:</b> John Oliver Clip  <b>Watch:</b> retort to Oliver Clip  <b>Read:</b> 2016 Bias and pain, PNAS</p>