

# Human Molecular Genetics

## BI 423/523 W2022

**Lecture Time:** 10:00-11:20 Monday & Wednesday

**Lecture Location:** 131 GSH (this is an in-person lecture)

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

**Office Hours will be held via zoom to maximize access:** R 10-11 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, links on [canvas homepage](#).
2. This course uses a **Teams** site for shared work, all students have UO granted access to Teams <https://service.uoregon.edu/TDClient/2030/Portal/KB/ArticleDet?ID=100604> you can access teams through your web browser, you don't need to download it. Your project content will be uploaded to teams so that we can all share each-others work.
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. Please bring your computer or tablet to lecture with you.
6. Posting course material including videos, presentations, infographics, and assessments on any platform that is not officially affiliated with the course is prohibited and will be treated as academic misconduct and reported to the Dean of Students Office.

### 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 grade for this course will be based on homework assessments (56 points) and 4 group projects (44 points) that include an individual written component and a group presentation component. The projects will require you to investigate, in detail, one aspect of a much broader genetics topic. You can expect to spend approximately 10-15 hours per project on doing research and preparing materials for the presentations (20 hours for graduate students). You can expect to spend approximately 5-10 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**

4 projects: Each project will be worth 11 points (44pt total), these include:

Short presentation (about 4 min)

Infographic (one page)

At least 2, well justified and properly cited, references

Vocab list for topic

Working well together within and outside of class

Self/group assessment

Specific expectations for each component can be found on Canvas at the top of the homepage

The remaining 58 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 quizzes at the start of each class (for which these is assigned content).

## **Absence policy:**

If you are sick, please stay home and take care of yourself. If you have been asked to quarantine, you should also not come to class. Aside from these two cases, attendance in class is mandatory; much of the class content will be discussed and presented in groups during class.

## **Makeup work:**

- preparatory work assessment:

If you are not in class for the preparatory work assessment you can make up the credit by submitting a two page, single spaced, 12 point font, summary of the assigned content. This needs to be in essay form and in your own words and include your thoughts on the content. You will also need to come to zoom office hours to discuss the content, unless you are too ill to zoom, in that case we will figure something out for when you are better.

- Presentation:

If you cannot make it to the presentation day you can record your presentation using the PowerPoint record option, or using Zoom. Please let me know if you are not sure how to do this. Your recording should be uploaded to our teams site. You will also need to come to zoom office hours to discuss the content, unless you are too ill to zoom, in that case we will figure something out for when you are better.

## Course/University policies <https://provost.uoregon.edu/syllabus-guidelines>

### Academic Disruption due to Campus Emergency

“In the event of a campus emergency that disrupts academic activities, course requirements, deadlines, and grading percentages are subject to change. Information about changes in this course will be communicated as soon as possible by email, and on Canvas. If we are not able to meet face-to-face, students should immediately log onto Canvas and read any announcements and/or access alternative assignments. Students are also expected to continue coursework as outlined in this syllabus or other instructions on Canvas.

In the event that the instructor of this course has to quarantine, this course may be taught online during that time.”

### COVID Containment Plan for Classes

As the University of Oregon returns to in-person instruction, the key to keeping our community healthy and safe involves **prevention, containment, and support**. Here is information critical to how the UO is responding to COVID-19.

- **Prevention:** To prevent or reduce the spread of COVID-19 in classrooms and on campus, all students and employees:
  1. Must comply with [vaccination policy](#)
  2. Must [wear face coverings](#) in all indoor spaces on UO campus
  3. Complete weekly [testing](#) if not fully vaccinated or exempted
  4. [Wash hands](#) frequently and practice social distancing when possible
  5. Complete daily [self-checks](#)
  6. Stay home/do not come to campus if feeling [symptomatic](#)
  7. Complete the UO [COVID-19 case and contact reporting form](#) if you test positive or have been in close contact with a confirmed or presumptive case.
- **Containment:** If a student in class tests positive for COVID-19, all relevant classes will be notified via an email by the Corona Corps Care Team with instructions for students and staff based on their vaccination status. Specifically:
  - **Vaccinated and Asymptomatic students:** Quarantine not required, but daily self-monitoring before coming on campus is advised; sign up for testing through MAP 3-5 days after exposure if advised you are a contact.”
  - **Unvaccinated or partially vaccinated students:** 14-day quarantine advised – do not come to class – and sign up for testing 3-5 days after notification through [MAP](#), if asymptomatic, or through University Health Services (541-346-2770) or your primary care provider, if symptomatic.
  - **Symptomatic students:** stay home (do not come to class/campus), complete the online [case and contact form](#), and contact University Health Services (541-346-2770) or your primary care provide to arrange for immediate COVID-19 testing.

Students identified as a **close contacts** of a positive case will be contacted by the Corona Corps Care Team (541-346-2292).

- **Support:** The following resources are available to you as a student.
  - [University Health Services](#) or call (541) 346-2770
  - [University Counseling Center](#) or call (541) 346-3277 or (541) 346-3227 (after hrs.)
  - [MAP Covid-19 Testing](#)
  - [Corona Corps](#) or call (541) 346-2292
  - [Academic Advising](#) or call (541) 346-3211
  - [Dean of Students](#) or call (541)-346-3216

### Good Classroom Citizenship

- Wear your **mask** and make sure it fits you well
- **Stay home** if you're sick
- **Get to know your neighbors** in class, and let them know if you test positive
- **Get tested** regularly
- Watch for **signs and symptoms** with the daily symptom self-check
- **Wash your hands** frequently or use hand sanitizer

Complete the UO COVID-19 [case and contact reporting form](#) if you test positive or are a close contact of someone who tests positive.”

**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](http://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](http://safe.uoregon.edu), [respect.uoregon.edu](http://respect.uoregon.edu), or [investigations.uoregon.edu](http://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](#).”*

**Mental Health and Wellness** – Life at college can be very complicated. Students often feel overwhelmed or stressed, experience anxiety or depression, struggle with relationships, or just need help navigating challenges in their life. If you're facing such challenges, you don't need to handle them on your own--there's help and support on campus. As your instructor if I believe you may need additional support, I will express my concerns, the reasons for them, and refer you to resources that might be helpful. It is not my intention to know the details of what might be bothering you, but simply to let you know I care and that help is available. Getting help is a courageous thing to do—for yourself and those you care about.

University Health Services help students cope with difficult emotions and life stressors. If you need general resources on coping with stress or want to talk with another student who has been in the same place as you, visit the Duck Nest (located in the EMU on the ground floor) and get help from one of the specially trained Peer Wellness Advocates. Find out more at [health.uoregon.edu/ducknest](http://health.uoregon.edu/ducknest).

University Counseling Services (UCS) has a team of dedicated staff members to support you with your concerns, many of whom can provide identity-based support. All clinical services are free and confidential. Find out more at [counseling.uoregon.edu](http://counseling.uoregon.edu) or by calling 541-346-3227 (anytime UCS is closed, the After-Hours Support and Crisis Line is available by calling this same number).

**Basic Needs** – Any student who has difficulty affording groceries or accessing sufficient food to eat every day, or who lacks a safe and stable place to live and believes this may affect their performance in the course is urged to contact the Dean of Students Office (346-3216, 164 Oregon Hall) for support.

This UO webpage includes resources for food, housing, healthcare, childcare, transportation, technology, finances, and legal support: <https://blogs.uoregon.edu/basicneeds/food/>

**Accommodation for Religious Observances** – The university makes reasonable accommodations, upon request, for students who are unable to attend a class for religious obligations or observance reasons, in accordance with the university discrimination policy which says “Any student who, because of religious beliefs, is unable to attend classes on a particular day shall be excused from attendance requirements and from any examination or other assignment on that day. The student shall make up the examination or other assignment missed because of the absence.” To request accommodations for this course for religious observance, visit the Office of the Registrar's website (<https://registrar.uoregon.edu/calendars/religious-observances>) and complete and submit to the instructor the “Student Religious Accommodation Request” form prior to the end of the second week of the term.

Course schedule and assignments (subject to change)

Mon January	Wed
3 Intro to class Read: <a href="#">2008 Genetic Research in Native Communities</a> set up: <a href="#">UO VPN account</a>	5 High throughput sequencing (HTS) Watch: <a href="#">Sequencing video from BI 320</a> Listen: <a href="#">2019 NPR story: HTS in medical diagnostics</a> Read: : <a href="#">2015 HTS review, Molecular Cell (presentation paper)</a> Optional iBiology videos Eric Chow: <a href="#">1</a> <a href="#">2</a> <a href="#">3</a> <a href="#">4</a> <a href="#">5</a> (5 talks about single cell sequencing)
10 HTS presentations	12 Human Genome Watch: <a href="#">Genome organization video from BI 320</a> Listen: <a href="#">2019 NPR story: genetic surveillance</a> Read: <a href="#">2019 Genomic surveillance, Nature</a> Read: <a href="#">2016 Genomic diversity, Nature</a> Read: <a href="#">2014 Functional DNA, PNAS (optional supplement)</a>
17 MLK <a href="#">Memorable speeches PBS</a>	19 CRISPR Cas9 Gene editing Watch: <a href="#">CRISPR Cas9 video from BI 320</a> Watch: <a href="#">Daudna talk</a> Read: web: <a href="#">How Does Genome Editing Work?</a> Read: <a href="#">2018 CRISPR Cas9, Nature Communications (Pres paper)</a>
24 Prime editing Watch: <a href="#">Kevin Esvelt iBiology Gene Drive Video</a> Read: <a href="#">2019 Prime Editing, Science</a> Read: <a href="#">2019 Prime Editing, Nature</a>	26 Gene editing and ethics Watch: <a href="#">John Oliver (skip the mouse/tick part)</a> Watch: <a href="#">Video on Zolgensma site</a> Listen: <a href="#">2019 Zolgensma NPR</a> Listen: <a href="#">2018 NPR story: gene-edited babies</a> Read: <a href="#">2019 BBC update on gene-edited babies</a> Read: <a href="#">2017 Gene Therapy, A Threat to the Deaf Community?</a> Read (might be too much, lets discuss): <a href="#">2018 Gene Therapy, Science</a>
Mon February	Wed
31 Editing presentations	2 Chromatin Structure, Topologically Associating Domains (TADs) Watch: <a href="#">2019 part 1 &amp; 2 iBiology cohesin &amp; CTCF</a> Read: <a href="#">2019 Genome folding into TADs, Science Advances (Presentation paper)</a>
7 Structure and disease Read: : <a href="#">2019, Crom Topology in devo and disease, Curr Opin Gen and Dev</a>	9 TADs Loop Extrusion Read: <a href="#">2018, Imaging loop extrusion, Science (optional supplement)</a> Watch: videos for 2018 paper: <a href="#">loop ext disruption</a> , <a href="#">loop ext no flow</a> , <a href="#">loop ext with flow</a> Read: <a href="#">2019, loop extrusion by human cohesin, Science</a>
14 Structure presentations	16 non-coding RNA (ncRNA) Read: <a href="#">2014 Rise of Regulatory RNAs, Nat Rev Gen (Presentation paper)</a>
21 X inactivation Watch: <a href="#">2018 iBiology 3 parts</a> Read: <a href="#">2011 Reg of X inactivation, Nat Rev Gen</a>	23 X inactivation Read: <a href="#">2015 X inactivation, Nature</a>
Mon March	Wed
28 ncRNA presentations	2 Splicing Read: <a href="#">2017 Targeting splicing in human disease, Genes</a>
7 Spinraza Read: <a href="#">2017 Spinraza, Nat Neuro</a> Read: <a href="#">2017 Spinraza, Transl Neuro</a>	9 Bias and pain Watch: <a href="#">John Oliver Clip</a> Watch: <a href="#">retort to Oliver Clip</a> Read: <a href="#">2016 Bias and pain, PNAS</a>