# Molecular Genetics and Genomics

## University of Victoria, BIOL 361

## Spring 2024

***Instructors***: Dr. Ryan Gawryluk — ryangawryluk@uvic.ca
Dr. Jürgen Ehlting — je@uvic.ca

***Lectures***: 8:30 to 9:50 am Mondays and Thursdays in Elliot Building 167

***Office hours***: By appointment (in person or through Zoom),
inquire with the lecture instructor whom you would like to meet with.

### *Course Description:*

Naturally occurring and induced genetic mutations leading to phenotypic variation within- and among-species in a diversity of eukaryotic taxa, including yeasts, plants, and animals. Theory and mechanisms in regulation of transcription and translation, genome structure and mobile genetic elements, and functional genomics.

### *Intended Learning Outcomes:*

Students will gain a comprehensive understanding of genomics from foundational concepts to advanced technologies and their applications across different research areas including health, evolution, and biotechnology. Students learn to integrate various concepts, technologies, and methodologies into a comprehensive understanding of genomics.

Specifically, by the end of the course students will understand:

- *Genetic Mapping Techniques*: They will understand how genetic mapping and genome-wide association approaches can unveil the molecular mechanisms underpinning animal phenotypic variation and human disease.
- *Genomic Technologies*: They will gain familiarity with genomic technologies such as whole genome sequencing and assembly, metagenomics, and transcriptomics.
- *Genome Evolution and Structural Variations*: They will understand the impact of transposable elements on genome evolution, gene/genome duplication mechanisms, and the plasticity of genome structures.
- *Functional Genomics*: They will explore the mechanisms of transcription, translation, and gene regulation with a focus on the roles that non-coding RNAs have in these processes.
- *Aspects of Applied Genomics*: Students will gain understanding of applied topics such as synthetic biology, CRISPR-based genome editing, cancer genomics, plant transgenesis, and phylogenetics through immersion in application-based sessions.

***Resources:***

Short summaries, lecture slides, and complete references of key papers will be uploaded to BrightSpace. The text *Molecular Biology of the Cell* by Alberts et al. (the textbook also used in BIOL360) will be very useful but is not required.

***Attendance:***

This is an in-person course but we encourage you to *stay home if you have symptoms of any communicable disease (cold, flu, COVID,…).* All lectures will be recorded (unless technical problems impede recordings) and made available on BrightSpace. Participation activities will also be available remotely.

***Assessment:***

One hundred (100) marks in total:

There will be ***three exams***, each worth 27 marks, for a total of 81 marks. All exams are ***non-cumulative*** and will only cover the course material since the previous exam. They will all have the same length. The first two exams will take place during class time, while the third exam will take place during the scheduled 'final exam' time slot (date to be determined). There will be ***one group assignment*** worth 16 marks. Detailed assignment instructions will be handed out in early February. It will involve reading primary literature and summarizing how genomic technologies have facilitated understanding of a topic in biology.

There will be 3 marks for completing three short ***assigned 'reflections' about selected topics*** in the class prior to exams (3 marks). This is a participation mark that will not be evaluated.

*Notes on exams:*

All exams will take place ***during regular lecture time***, likely via BrightSpace. If you do not have a device you can use to write the exam on BrightSpace, please contact us as at the beginning of the course to guide you to resources (e.g. the 'Borrow a Laptop' program from the library). Exams are designed for a ***duration of 50 minutes*** starting at the beginning of the lecture time slot, and there will be a hard end time by which you have to submit your exams. Ad hoc extensions will only be granted if technical problems arise, for example if the connection to the BrightSpace server got interrupted. Students must inform invigilators immediately about lost internet connections. If you are fit to write the exam but should not come to campus (e.g., to prevent the spread of communicable diseases), please contact the instructors as early as possible to discuss options.

Exams are ***closed-book and invigilated***, but you are allowed to bring to the exams a **'cheat-sheet'**, i.e. a single sheet of paper with as many handwritten notes on both sides as you can make fit. Note however, that exams are timed and that you will not have sufficient time during the exam to extensively evaluate the material you brought in.

There will be ***no supplemental in-course exams***. Please inform the course coordinator as soon as possible (within one week), if you had to miss an exam due to unexpected and unavoidable circumstances, or conflicting responsibilities (as detailed here). In this case the evaluation breakdown will be adjusted accordingly, that is the remaining two exams will each be worth 40.5% of your final grade. Exams missed without excuse will be given zero marks. ***Writing at least two exams is a course requirement***. Failure to do so will result in a grade of "N" regardless of the cumulative percentage on other elements of the course. Remember that N is a failing grade and factors into GPA as a value of zero. Students that obtained a grade of "N" may submit a formal Request for Academic Concession (RAC) and a deferred exam covering the missed course material will be organized if the RAC has

been approved.

***Academic Integrity Statement***

Students must abide by academic regulations as set out in the university calendar. They must observe standards of scholarly integrity with regards to plagiarism and cheating. Please read the definitions, watch the tragi-comic video, and look at the other information available at this link.

The use of generative AI tools, such as ChatGPT or GrammarlyGO, are not allowed for assignments (or during closed book exams, of course). We reserve the right to use anti-plagiarism or AI detection software.

***Grading scheme***

A+ (90%-100%), A (85-89.75%), A- (80-84.75%), B+ (77-79.75, B (73- 76.75%),

B- (70-72.75%), C+ (65-69.75%), C (60-64.75%), D (50-59.75%), F (<50%)

***Academic Important Dates:***

Check here. It is the student's responsibility to attend to Add/Drop dates published in the Calendar (last day to add courses – Jan 24, last day to drop without failure – Feb 29). Students must not assume they will be dropped automatically from any course they do not attend. It is also the students' responsibility to check their records and registration status.

## *Tentative course schedule*

**Week 1** (Jan 8- 12)

Jan 8: Introduction to the course and outline of material [JE, short intro by RG]

Jan 11: Qualitative and quantitative genetic loci: Sticklebacks with a no-pelvis phenotypes. [JE]

**Week 2** (Jan 15 - 219)

Jan 15: Mapping Pitx1: the transcription factor responsible for pelvis phenotypic variation [JE]

Jan 18: Molecular basis of convergent evolution of hind-limb reduction in animals. [JE]

**Week 3** (Jan 22 - 26)

Jan 22: SNP arrays, mapping, and human diseases: Clubfoot disease. [JE]

Jan 25: Genome wide association genetics and human diseases: Anemia, BCL11 and hemoglobin. [JE]

**Section 1: Phenotype to Genotype**

- Stickleback pelvic fins and genetic mapping
- Genome wide association genetics and human diseases → Markers, mapping, and genetic associations

***Intended Learning Outcomes:***

throughout this section you will

- differentiate genetic mapping techniques (from classic genetic mapping to genome wide association studies)
- distinguish different mechanisms how genetic variation can cause phenotypic variation.
- recognize how genomics association studies can help understanding human diseases.

**Week 4** (Jan 29 – Feb 02)

Jan 29: Whole genome sequencing technology and methods. [JE]

Feb 01: Putting it all together, how to assemble a whole genome. [JE]

**Week 5** (Feb 05 - 09)

Assignment handed out.

Feb 05: Eukaryotic genomes – an introduction to transposable elements [RG]

Feb 08: **Exam 1** [JE/RG]

**Week 6** (Feb 12 - 16)

Feb 12: Transposable elements shape genomes and influence evolution I [RG]

Feb 15: Transposable elements shape genomes and influence evolution II [RG]

**Week 7** (Feb 19 – 23)

Reading Break: no lectures

**Week 8** (Feb 26 – Feb 30)

Feb 26: Genome structure and plasticity: Gene/Genome duplication mechanisms [RG]

Feb 29: Diverse functions of RNAs in transcription and translation [RG]

**Week 9** (Mar 04 - 08)

Mar 04: The discovery of small regulatory RNAs in *Caenorhabditis elegans* [RG]

Mar 07: Post-transcriptional regulation of gene expression via small interfering RNAs [RG]

**Week 10** (Mar 11 – 15)

Mar 10: Mechanisms of small RNA regulatory action [RG]

Mar 14: Programmed genome elimination [RG]

**Week 11** (Mar 18 - 22)

Mar 18: **Exam 2** [JE/RG]

Mar 21: CRISPR and genome editing [RG]

**Section 2: Genome Structure and Plasticity**

- Whole genome sequencing, and genome structure
- Transposable elements and their impact on evolution
- Gene and genome duplications

***Intended Learning Outcomes:***

throughout this section you will

- understand how DNA sequencing technologies evolved and enable whole genome sequencing
- appreciate key structural elements that make up eukaryotic genomes and how they can facilitate evolutionary change

**Section 3: The diverse functions of non-coding RNAs**

- The diverse functions of RNAs beyond being a messenger
- Small regulatory RNAs and mechanisms of small RNA regulatory action
- Genome elimination mechanisms
- RNAs in defense against viruses: CRISPR/Cas

***Intended Learning Outcomes:***

throughout this section you will

- recognize that RNAs have informational, structural, enzymatic, and regulatory functions related to realizing gene expression
- understand that RNAs evolved to acquire function in shaping genome structure and in defense against 'molecular parasites'

**Week 12** (Mar 25 – 29)

Mar 25: The cancer microbiome [RG]

Mar 28: Mammalian tumors and their causes: alterations of cancer-critical genes [JE]

**Week 13** (Apr 01 – 05)

Apr 01: Easter Monday, no lecture

Apr 04: Genomics (and transcriptomics) of mammalian tumor formation [JE]

**Week 14** (Apr 08 - 12)

Apr 08: Plant tumors / trangenics [JE] (if time allows)

**Exam 3**: In scheduled exam slot [JE/RG]

**Section 4: Cancer formation in mammals and plants**

- Cancer and signal transduction pathways
- Genomics of mammalian cancer formation
- Tumor formation in plants (Inter-kingdom lateral gene transfer, transgenic plants)

***Intended Learning Outcomes:***

throughout this section you will

- review the basic molecular mechanisms causing mammalian cancer formation
- recognize how high-throughput sequencing revolutionizes cancer research towards personalized treatment
- grasp the fundamental differences in tumor formation in plants and animals

***Land Acknowledgement:***

We acknowledge and respect the lək̓ʷəŋən peoples on whose traditional territory the University of Victoria stands, and the Songhees, Esquimalt and W̱SÁNEĆ peoples whose historical relationships with the land continue to this day. We are thankful to be able to learn together on this land and strive to make the world a better place.

***Diversity, Inclusion, and Resources*:**

We welcome everyone to learn in this course and we respect every human being, from all ethnic backgrounds, religious beliefs, sexual orientations, genders, socio-economic backgrounds and abilities. UVic is committed to promoting, providing and protecting a supportive and safe learning and working environments for all of its members. Your health and mental well-being are important for succeeding in this course, so please take advantage of UVic resources:

**Centre for Accessible Learning** - The CAL staff are available by appointment to assess specific needs, provide referrals and arrange appropriate accommodations. Accommodation letters need to be released to instructors at least seven days in advance of an assessment for accommodations to be applied. Please meet with the instructor(s) about specific concerns if needed.

**Learn Anywhere** – provides extensive information and student supports for online technology. This portal helps navigate essential resources, services, learning tools and strategies.

**Student Mental Health Supports -** In addition to providing both face to face and online mental health resources through the new Student Wellness Centre, a 24 x 7 phone & online student mental health resource and support program (Support Connect) is available for all UVic students, no matter where you are located, at any time.

**Student Wellness** - provides a full service, primary health clinic for students, and coordinates healthy student and campus initiatives. They also offer counselling services that can help you make the most of your university experience. They offer free professional, confidential, inclusive support to currently registered UVic students.

**Elders' Voices -** The Office of Indigenous Academic and Community Engagement (IACE) has the privilege of assembling a group of Elders from local communities to guide students and others in

Indigenous ways of knowing and being.

**Office of Student life** - promoting a safer and more inclusive campus community and supporting student engagement, development and well-being

**Sexualized Violence Prevention and Response** - UVic takes sexualized violence seriously. We encourage students to learn more about how the university defines sexualized violence and its overall approach. If you or someone you know has been impacted by sexualized violence and needs information, advice, and/or support please contact the sexualized violence resource office in Equity and Human Rights (EQHR). If you want to take part in the important prevention work taking place on campus, you can also contact the sexualized violence resource office in EQHR; Sedgewick C119, Phone: 250.721.8021, Email: svpcoordinator@uvic.ca.