

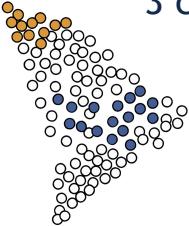


School of Molecular & Cellular Biology

MCB 545, Spring 2026

Functional Genomics in Principle and Practice

3 credit hours



Instructor

Kevin Van Bortle

B521 CLSL

kvbortle@illinois.edu

Class Meeting Schedule

M,W 1:30pm - 2:50pm

Location: Nevada Building Computer Lab (1203 ½ W. Nevada St.)

Office Hours: Fridays, 2:00 - 3:00 pm (virtual)

Course Overview and Description

This course introduces both the experimental and analytical foundations of functional genomics tailored to students interested in understanding and leveraging modern genomic data to analyze gene regulation and function. Lectures cover basic principles (genome assembly, annotation, transcriptomics and epigenomics, functional perturbation methods, statistics and significance testing for genomic data, etc.) and are combined with hands-on programming and genomic data analysis. Students will apply concepts learned through lecture and discussion to the actual study of RNA-binding proteins, including gene regulatory mechanisms and downstream RBP function, by analyzing public, multi-omic data generated by recent genomic consortia, including ENCODE, The 4D Nucleome (4DN), and The Cancer Genome Atlas (TCGA). Students submit a final written synthesis of their findings and underlying code at the conclusion of the course.

Course Prerequisites

Students are encouraged to familiarize themselves with RStudio and the basics of programming in R prior to taking this class.

Student Learning Outcomes

At the end of the course, through assignments, discussions, activities and assessments, students will be able to:

- Understand basic principles of modern functional genomics methods, including current sequencing, computational, and statistical approaches
- Interpret common data analysis and visualization techniques and apply these approaches using programming in R
- Integrate multi-omic data to generate quantitative analyses related to the regulation and function of a specific gene or gene set of interest.

Course Text/Materials Information

Primary literature and reviews related to lecture topics will be shared over the course of the semester. Students are advised to become familiar with programming in R prior and/or in the first few weeks of the semester (example resource below). Student exercises and assignments will include a series of R markdown files provided in class.

Hands-on programming with R. Garrett Grolemund. O'Reilly Media. 2014
<https://rstudio-education.github.io/hopr/basics.html>

Course Tools

MCB545 will utilize a Slack workspace for course communication between instructor and students (<https://slack.com>). Course material will be shared through Slack; data resource files will be accessed via GitHub (https://github.com/kvbortle/MCB545_FunctionalGenomics)

Grading Information and Breakdown

15% Class participation: includes attendance and active participation (15 points total)

40% Graded homework assignments (40 points total, 10 points each)

15% Graded Midterm Exam (15 points total)

10% Final class presentation (RBP genomic analyses and findings; 10 points total)

20% Final Rmarkdown report (RBP analyses, interpretation, and code; 20 points total)

100 Total points

Grade format

97-100% (A+)	93-96% (A)	90-92% (A-)
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87-89% (B+)	83-86% (B)	80-82% (B-)
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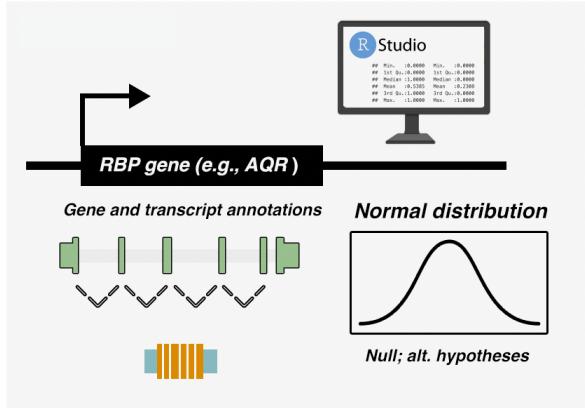
77-79% (C+)	73-76% (C)	70-72% (C-)
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67-69% (D+)	63-66% (D)	60-62% (D-)
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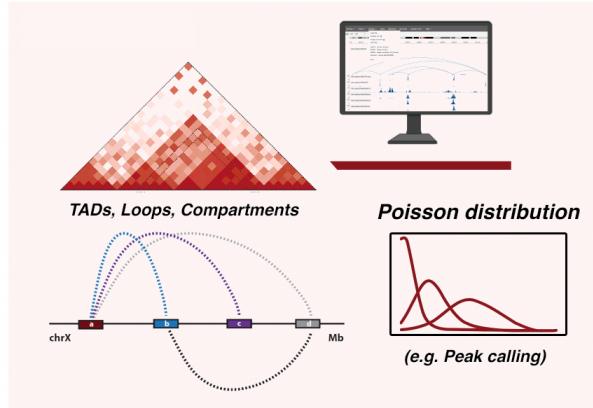
0-59% (F)		
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VISUAL COURSE OUTLINE (2026)

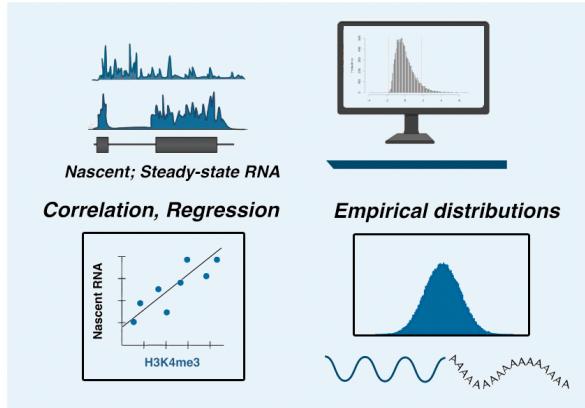
Module I. Intro to genomics and programming in R



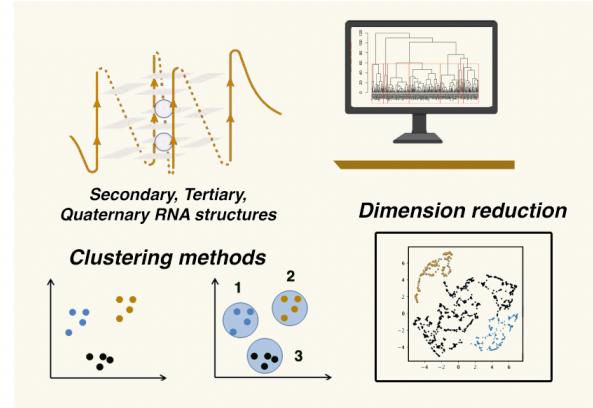
Module II. Chromatin architecture and regulation



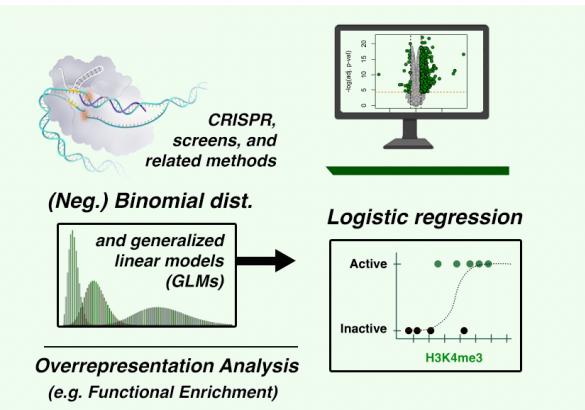
Module III. Transcription and RNA processing



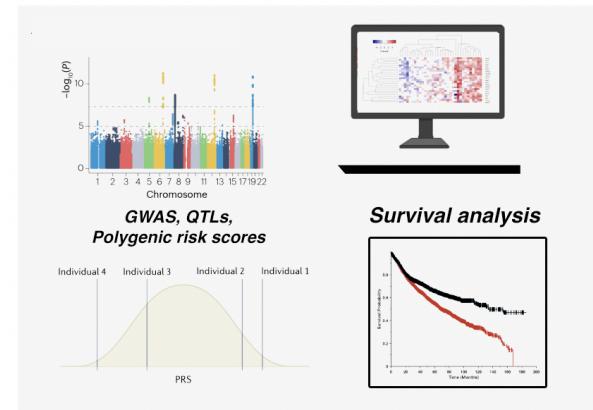
Module IV. RNA structure and translation



Module V. Perturbation and differential analysis



Module VI. Special topics (Health, disease, careers)



| COURSE CALENDAR (2026) |

SECTION I. INTRODUCTION TO GENOMICS | INTRO TO PROGRAMMING IN R

January 21	<u>Genomics Topic:</u> Course overview Intro to genomics RBP selection <u>Week 1 Reading:</u> https://rstudio-education.github.io/hopr/basics.html <u>Assignment:</u> Intro Poll; RNA-binding protein selection; install Rstudio
January 26	<u>Genomics Topic:</u> Genome Assembly Gene Annotation Intro to IGV <u>Week 2 Reading:</u> Rood & Regev 2021 Mudge and Harrow, 2016
January 28	<u>Exercise:</u> Programming in R (Importing data, R functions, Plotting)
February 2	<u>Genomics Topic:</u> Sequencing Platforms Read Alignment <u>Week 3 Reading:</u> Shendure et al., 2017 Sahlin et al., 2023
February 4	<u>Exercise:</u> For Loops and If Statements in R Transcript feature survey <u>QMA Concept:</u> Empirical, Theoretical Distributions; Significance testing

Assignment # 1 - RBP gene and transcript annotation features: Due Feb. 13 (10 pts)
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SECTION II. CHROMATIN ARCHITECTURE AND REGULATION (IT'S A DNA WORLD)

February 9	<u>Genomics Topic:</u> Chromatin, and Transcription Factor (TF) Mapping <u>Week 4 Reading:</u> Klemm 2021 Meyer & Liu 2014
February 11	<u>Exercise:</u> Surveying the Regulome (Histones, Enhancers, Loops) <u>QMA Concept:</u> Poisson Distribution for peak calling, MHT correction
February 16	<u>Genomics Topic:</u> Loops, Domains, Compartments, and Replication <u>Week 5 Reading:</u> Szalay et al., 2024 Kempfer & Pombo, 2020 Jerkovic & Cavalli, 2021 Hu & Stillman, 2023
February 18	<u>Exercise:</u> Working w/ Genomic intervals ex: Visualizing chromatin state <u>QMA Concept:</u> Hidden Markov Models (HMM) for chromatin state
February 23	<u>Genomics Topic:</u> Location: Subnuclear position (e.g. LADs, Speckles) <u>Week 6 Reading:</u> Shan et al., 2024 Bhat et al., 2021
February 25	<u>Exercise:</u> Working with Genomic Intervals example 2: Nuclear Position <u>QMA Concept:</u> Permutation Tests with Genomic Data

Assignment # 2 - DNA-related features of your RBP-encoding gene: Due Mar. 6 (10 pts)
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SECTION III. TRANSCRIPTION AND RNA PROCESSING (FROM DNA TO RNA)

March 2	<u>Genomics Topic:</u> Transcription RNA-seq methods <u>Week 7 Reading:</u> <i>Stark et al., 2019 Wissink et al., 2019</i>
March 4	<u>Exercise:</u> Integrating Gene Expression and Chromatin Data <u>QMA Concept:</u> Linear Regression, Correlation Analysis
March 9	<u>Genomics Topic:</u> RNA processing and Splicing <u>Week 8 Reading:</u> <i>Neil et al., 2022; Rogalska, Vivori, Valcarcel 2023; Mitschka & Mayr, 2022; Childs-Disney et al., 2022</i>
March 11	MIDTERM EXAM (15 pts)

March 14 - 22 Spring Break

SECTION IV. RNA STRUCTURE AND TRANSLATION (FROM RNA TO PROTEIN)

March 23	<u>Genomics Topic:</u> RNA structure and Translation <u>Week 9 Reading:</u> <i>Brar & Weissman, 2015 Greener et al., 2022</i>
March 26	<u>Exercise:</u> mRNA G4 structures and splicing survey <u>QMA Concept:</u> Permutations Revisited (Intron ratios)
March 30	<u>Genomics Topic:</u> Protein-RNA & RNA-RNA Interaction Mapping <u>Week 10 Reading:</u> <i>Hafner et al., 2021 (CLIP and complement. methods)</i>
April 1	<u>Exercise:</u> RNA-binding preferences and target RNA populations <u>QMA Concept:</u> Dimension Reduction (PCA and other Techniques)

| Assignment # 3 - RNA-related features of your RBP-encoding transcript: Due Apr. 10 (10 pts) |

SECTION V. PERTURBATION AND DIFFERENTIAL ANALYSIS (PROTEIN “FUNCTION”)

April 6	<u>Genomics Topic:</u> Functional Genomics; Gene Modulation (e.g. CRISPR) <u>Week 11 Reading:</u> <i>Dowdy, 2017; Adli, 2018</i>
April 8	Exercise: Introduction to Linux, Biocluster, and HPC (retrieval of eCLIP, differential expression, and splicing data)
April 13	<u>Genomics Topic:</u> Differential Analysis Functional Enrichment (intro) <u>Week 12 Reading:</u> <i>Van den Berge et al., 2019</i>
April 15	Week 13 Exercise: Gene Set Enrichment Analysis <u>QMA Concept:</u> Fisher’s Exact Test for overlap enrichment
April 20	<u>Genomics Topic:</u> Functional Enrichment (cnt’d) Classification Mths. <u>Week 13 Reading:</u> <i>Garcia-Campos et al., 2015 Mubeen et al., 2022 Wijesooriya et al., 2022 Bild et al., 2006</i>
April 22	Week 13 Exercise: Integrating RBP eCLIP and differential genes <u>QMA Concept:</u> Logistic Regression for modeling binary outcomes

| Assignment # 4 - RBP-binding and target RNA regulatory prediction: Due May 1 (10 pts) |

SECTION VI. SPECIAL TOPICS (HEALTH, DISEASE, CAREERS); PRESENTATIONS

April 27	<u>Genomics Topic:</u> Genomics in Health and Disease <u>Week 14 Reading:</u> <i>Malik et al., 2021; Uffelmann et al., 2021</i>
April 29	Exercise: Is my RBP a prognostic factor in cancer? <u>QMA Concept:</u> Survival Analysis
May 4 & May 6	PRESENTATIONS (10 pts)

Special “Careers in Genomics” Seminar TBA

| FINAL REPORT DUE MAY 13 (20 pts) |

Assignments, due dates, and course expectations

MCB545 is geared towards the practical application of functional genomic data analysis and will function as a hybrid lecture and hands-on lab course series. Attendance and in-class participation (maximum 15 points, 15% of total) is critical for actively learning this broad subject area. Students must contact me immediately or in advance if it becomes necessary to be absent from class to identify a reasonable action plan. Lecture topics will be linked to learning programming in-class and through multiple homework assignments that will be submitted electronically (maximum 40 points, 40% of total). This course encourages team-learning, including regular communication and group work on the MCB545 Slack workspace. However, homework assignments must be written up and submitted independently. A mid-semester in-class written exam will be used to assess learning outcomes (maximum 15 points, 15% of total) and, towards the completion of the semester, a final presentation (maximum 10 points, 10% of total) and project assignment (maximum 20 points, 20% of total) will require each student to independently apply data processing tools and data analysis methods taught during the semester, as well as integrate and synthesize all data analyzed throughout the course to establish a hypothesis about the regulation and function of a specific gene.

Academic integrity

Students are expected to be familiar with the code of policies and regulations applied in all instances of academic misconduct. Please refer to <http://studentcode.illinois.edu>, and, in particular, Article 1 part 4: <http://studentcode.illinois.edu/article1/part4/1-401/>

Accommodations

To obtain disability-related academic adjustments and/or auxiliary aids, students with disabilities must contact the course instructor and the Disability Resources and Educational Services (DRES) as soon as possible. To contact DRES, you may visit 1207 S. Oak St., Champaign, call 333-4603 (V/TDD), or e-mail a message to disability@uiuc.edu. <http://www.disability.illinois.edu/>.

Inclusive classroom statement

The effectiveness of this course is dependent upon the creation of an encouraging and safe classroom environment. Exclusionary, offensive or harmful speech, such as racism, sexism, homophobia, and transphobia, will not be tolerated and in some cases will be subject to University harassment procedures. We are all responsible for creating a positive and safe environment that allows all students equal respect and comfort. We expect each of you to help establish and maintain an environment where you and your peers can contribute without fear of ridicule or intolerant or offensive language.

Sexual misconduct policy and reporting

The University of Illinois is committed to combating sexual misconduct. Faculty and staff members are required to report any instances of sexual misconduct to the University's Title IX and Disability Office. In turn, an individual with the Title IX and Disability Office will provide information about rights and options, including accommodations, support services, the campus disciplinary process, and law enforcement options. A list of the designated University employees who, as counselors, confidential advisors, and medical professionals, do not have this reporting responsibility and can maintain confidentiality, can be found here: wecare.illinois.edu/resources/students/#confidential. Other information about resources and reporting is available here: wecare.illinois.edu.