# BINF 739 Topics in Bioinformatics:
# Next-generation Sequencing
# Fall 2022

**Meeting time:** Wednesdays 4:30 – 7:10 pm
**Meeting place:** Online through Blackboard Collaborate Ultra
**Credit hours:** 3.0 credits

**Instructor:** Dr. Amy Smith
**Office location:** Online
**Office hours:** By appointment
**Email:** asmi6@gmu.edu

**Course description:**
This course will introduce students to the principles of next-generation sequencing (NGS) techniques and practices. It is designed in three parts to bring the student from active listener to active participant.

- **Part 1** presents a brief overview of NGS technologies and computational algorithms. The R software environment and UNIX command line interface are introduced and/or reviewed.
- **Part 2** explores recent examples of genome, exome, transcriptome, and epigenome studies with appropriate background concepts provided.
- **Part 3** provides a hands-on opportunity for a student to work with publicly available NGS datasets in a small independent project.

**Prerequisites:**

- **Recommended:** Basic knowledge in molecular biology, probability, and statistics or permission of instructor
- **Preferred:** Familiarity with a programming language.

**Required Reading:**
Students will be assigned journal articles available through the GMU Libraries.

**Learning Outcomes:**

1. Students will know how to use bioinformatics resources to access and make sense of the wealth of data provided by NGS technologies to answer biologically relevant questions in their areas of interest.
2. Students will become familiar with a typical NGS analysis pipeline including assessment of raw reads, alignment to a genome, and gene-centric analyses.
3. Students will understand how experimental design, data acquisition, and data analysis are applied to and achieved with NGS studies.

**Grading:**

10% Class participation
15% Homework 1 (due 9/28)
15% Homework 2 (due 10/19)
20% Journal Club article (one time during semester)
40% Final project (due 12/7)

| Letter Grade | Percentage |
|---|---|
| A+ | >100 |
| A | 90-100 |
| B+ | 88-89.99 |
| B | 80-87.99 |
| C+ | 78-79.99 |
| C | 70-77.99 |

**Class Schedule:**

**8/24 Lecture 1:**

Class schedule and expectations
Introduction to next generation sequencing – Part 1

**8/31 Lecture 2:**

Introduction to next generation sequencing – Part 2

**9/7 Lecture 3:**

NGS raw data formats and the SRA
Basic UNIX commands and R

**9/14 Lecture 4:**

Alignment of sequence reads to a reference genome
Galaxy
Assign Homework 1

**9/21 Lecture 5:**

*De novo* sequence assembly

**9/28 Lecture 6:**

Identification of variants
Homework 1 Due

**10/5 Lecture 7:**

Genome/exome analysis
Journal Club
Assign Homework 2

**10/12 Lecture 8:**

Transcriptome/RNA-seq

Journal Club

**10/19 Lecture 9:**
ChIP-seq and Regulatory Genomics
Homework 2 Due

**10/26 Lecture 10:**
Applications in Human Disease
Journal Club
Assign Final Project

**11/2 Lecture 11:**
Applications in Infectious Disease – Viral Genomics
Journal Club

**11/9 Lecture 12:**
Applications in Infectious Disease – Immune Response
Journal Club

**11/16 Lecture 13:**
Epigenomics
Emerging Technologies
Journal Club

**11/23: No class, Thanksgiving break**

**11/30: Lecture 14:**
Question & Answer for final projects

**12/7: Final project due**

**Journal Club:**
Each student will take one turn presenting a brief overview of a research article relevant to the previous week's lecture topic. Students will sign up for one Journal Club slot between 10/5 and 11/16. Further details will be provided separately.

**Policies**
Students are encouraged to discuss course content, labs, and similar activities with other students; however, all assignment submissions must contain only original, individually completed work, unless a different arrangement approved by the instructor.

Posting or sharing course content (e.g., recordings, exams, or anything not created by the student), using any non-electronic or electronic medium (e.g., web site) where it is accessible to someone other than the individual student is strictly prohibited without prior instructor's approval.

Students must follow the university policy for Responsible Use of Computing (seehttp://universitypolicy.gmu.edu/policies/responsible-use-of-computing/).

Students are responsible for the content of university communications sent to their Masone mail account and are required to activate their account and check it regularly. All communication from the university, college, school, and program will be sent to students solely through their Mason email account.

Students with disabilities who seek accommodations in a course must be registered with George Mason University Disability Services. Approved accommodations will begin at the time the written letter from Disability Services is received by the instructor (seehttps://ds.gmu.edu/).

The class provides inclusive and equitable learning environment. It is expected that students adhere to the George Mason University Honor Code as it relates to integrity regarding coursework and grades. The Honor Code reads as follows: “To promote a stronger sense of mutual responsibility, respect, trust, and fairness among all members of the George Mason University community and with the desire for greater academic and personal achievement, we, the student members of the University Community have set forth this: Student members of the George Mason University community pledge not to cheat, plagiarize, steal and/or lie in matters related to academic work.” More information about the Honor Code, including definitions of cheating, lying, and plagiarism, can be found at the Office of Academic Integrity website at http://oai.gmu.edu