**SYLLABUS: Genomic Analysis I (PED 5750 / PED 575)**

**Department Offering Course:** Pediatrics  
**Course Number:** PED 5750 (for students), PED 575 (noncredit for faculty/staff/comm. through CECE)  
**Course Title:** Genomic Analysis I  
**Pre-requisites:** None, course is designed for graduate students or highly motivated undergraduates  
**Credit hours:** 1.5  
**Meeting time/location:** Thursdays 2:30pm-3:45pm, Health Sciences Education Building (HSEB) 1750  
**Instructor:** Clint Mason, PhD  
417 Wakara Way, Room 3117  
Salt Lake City, UT 84112  
(801) 587-3633  
**Office Hours:** Wednesdays 10:00am – 11:00am (if parking, please park in the lot south/west of the 421/423 buildings, vehicles may be ticketed if parked in the lot south of the 417 building)  
**Class website:**  
[home.chpc.utah.edu/~mason/Teaching/Genomic_Analysis_I/Genomic_Analysis_I_index.htm](http://home.chpc.utah.edu/~mason/Teaching/Genomic_Analysis_I/Genomic_Analysis_I_index.htm)  
**Email:** clint.mason@hsc.utah.edu (please begin the email header with PED 5750 or PED 575)  

**Course Description:** Provides comprehensive instruction on the analysis of genomic data. An overview of basic statistics, study design, genomic technologies, and computational software will be provided in addition to current best practices in the analysis of genomic data. Genomic Analysis I will focus on analysis and detection of variants and mutations from next generation sequencing data (whole genome sequencing, whole exome sequencing, and targeted panel sequencing) as well as the analysis of genomic copy number data.  

**Course Objectives:**  
1) Students will become familiar with the various genomic technologies currently in use for identifying variants, mutations, and copy number aberrations  
2) Students will be able to correctly design a Next Generation Sequencing (NGS) study (WGS or WES) and identify potential limitations of incorrectly designed studies  
3) Students will learn current best practices for alignment, processing, annotation, and variant calling in NGS data  
4) Students will learn basic statistical tests, be able to identify which are most appropriate for various genomic analysis scenarios, and apply them to assess the probability of specific NGS hypotheses  
5) Students will be able to process and analyze data from start-to-finish in a genomic analysis (NGS) project  

**Student requirements/evaluation criteria:** Students receiving a grade for the course are expected to attend class lectures and complete the weekly assignments. A final project to be completed and presented by each student near the end of the semester will comprise the major portion of the student’s grade. This project will require effort throughout the semester to complete. This is not a group project. While you may ask each other for advice, each student is responsible for running their own code and analysis by themselves and to present their work in the final presentation individually. Criteria for evaluation will include successful completion of the analysis project, use of appropriate code and statistics, and clarity in presentation of methods and results.
**Grade Scale:** A: 94-100%, A-: 90-93%, B+: 87-89%, B: 84-86%, B-: 80-83%, C+: 77-79%, C: 74-76%, C-: 70-73%, D: 60-69%, E: 0-59%

**Grade Composition:** Assignments will contribute 20% towards the final grade. The final personal project will contribute 80% towards the final grade.

**Teaching/learning methods:** This course will primarily be a lecture course with in-class discussion. Weekly assignments are meant to summarize the newly presented material of the week and ensure that the major concepts are understood by the student. The main learning by the student will occur through hands-on experience, trying out the code and concepts presented in class on real data sets, identifying problems in analyses, and finding solutions. Students are encouraged to explore their project as in-depth as possible, including attempting to generate results published in initial publications reporting the data. Doing and presenting what was done are considered to be the hallmark measures of learning in this class.

**Computational requirements/resources:** Each student may elect to receive temporary and restricted remote computational/storage space on the University of Utah’s CHPC resources if they understand and consent to the various requirements and regulations available at: home.chpc.utah.edu/~mason/agree/agree.htm. Students will still be required to have their own computer which can access the CHPC servers remotely. Students may instead obtain their own adequate personal computational resources and data (individual-level genomic data made available as a resource to students may not be downloaded to personal computers) to complete the project and assignments. However, this may involve a substantial amount of resources and such students should consult with the Instructor regarding necessary amounts.

**Restrictions on genomic analyses:** Students in this class are prohibited from attempting to personally identify individuals from any data set by their genomic data. If you are involved in such an effort through a project outside of this class you must inform the Instructor of such prior to the end of the first week of class. Various individual-level genomic data have been made available to the Instructor for use by consenting lab and class members who agree to abide by the various restrictions of the Database of Genotypes and Phenotypes (dbGaP). All regulations must be followed including the restriction to not download any of these individual-level genomic data off of the protected CHPC environment of the Instructor. The local University of Utah Institutional Review Board has approved the analyses to be performed in this class as non-human subjects research (IRB# 00088927).

**Copyright regulations:** Some or all material (including computer code, lecture notes, and in-class presentations) made available to students will be copyrighted and may not be copied, shared, posted, or forwarded to any other individual for any reason. Such resources if made available will be for the individual student’s personal learning benefit in this course only. Copyrighted code/software may not be used for outside-class projects without specific permission of the Instructor.

**Text/Reading lists:** Various online resources will be given throughout the class which explain various parts related to the course objectives. No textbook currently exists for this subject (utilizing the most current methods).

**Class Schedule:**

1/14 – Intro: genomics in 2016 vs 2011, genome biology/technologies, ethics, UofU computer resources
1/21 – Models, probability, statistics, and genomic study design
1/28 – Basic linux scripting, fastq data alignment to human genome, initial GATK steps
2/4 – GATK steps continued, variant output files (vcf, mpileup)
2/11 – Annotation and filtering of variant output files
2/18 – Germline variant detection and analysis
2/25 – De-novo mutation detection and analysis
3/3 – Somatic mutation detection and analysis
3/10 – Additional statistical assessments, graphical presentation of results
3/17 – (NO CLASS due to Spring Break)
3/24 – In depth in DNA sequence data (identification of sample mix-ups, shared heritance, etc.)
3/31 – Copy number estimation from NGS data
4/7 – Copy number estimation & SNP calling from microarray data
4/14 – Project presentations
4/21 – Project presentations, summary, collaboration projects

Assignment due dates: Assignments are due by the beginning of the following class (1-week completion time). The final class projects are due and to be presented on April 14th, 2016. Due to class size, some projects may be presented on April 21st, 2016 – but should still be completed by April 14th, 2016.

ADA statement: The Americans with Disabilities Act. The University of Utah seeks to provide equal access to its programs, services, and activities for people with disabilities. If you will need accommodations in this class, reasonable prior notice needs to be given to the Center for Disability Services, 162 Olpin Union Building, (801) 581-5020. CDS will work with you and the instructor to make arrangements for accommodations. All written information in this course can be made available in an alternative format with prior notification to the Center for Disability Services.

Title IX information: Title IX violations are encouraged to be reported to the Title IX Coordinator in the Office of Equal Opportunity and Affirmative Action, 135 Park Building, 801-581-8365, or the Office of the Dean of Students, 270 Union Building, 801-581-7066. For support and confidential consultation, contact the Center for Student Wellness, 426 SSB, 801-581-7776. To report to the police, contact the Department of Public Safety, 801-585-2677(COPS).

Academic code of conduct: Can be found at: http://regulations.utah.edu/academics/6-400.php

Faculty rights and responsibilities: Can be found at: http://regulations.utah.edu/academics/6-316.php

Accommodation policy: Can be found under Section Q: http://regulations.utah.edu/academics/6-100.php

Late/missed assignments policy: Late or missed assignments will have their credit reduced by 50%. No credit will be given for any assignments turned in following the last regularly scheduled class day of the semester.

Attendance policy: Regular attendance at all class meetings is expected.

Wellness resources: Personal concerns such as stress, anxiety, relationship difficulties, depression, cross-cultural differences, etc., can interfere with a student’s ability to succeed and thrive at the University of Utah. For helpful resources contact the Center for Student Wellness at www.wellness.utah.edu or 801-581-7776.

Changes to this Syllabus: Specific items in this syllabus may be modified or added to. Students will be informed of such changes during regularly scheduled class lectures.