TRANSLATIONAL BIOINFORMATICS

GMS 6804

3 CREDIT HOURS

SPRING 2017

LOCATION: HPNP Rm. 1101

MEETING TIMES: Monday and Wednesday 1-2.30pm

INSTRUCTOR: *François Modave, PhD, MS*
Office: 3217 Clinical and Translational Research Building
Email: modavefp@ufl.edu
Phone: (352) 294-5984

OFFICE HOURS: By appointment

COURSE COMMUNICATION: Students may email the instructor with questions, but are encouraged to consider whether their questions are of general interest to the entire class. Dedicated class time will be devoted to discussing and answering general questions about either course content or course mechanics that are relevant to all students.

MAIN TEXT (required)

1. Kann M, Lewitter F, eds. Translational Bioinformatics. Open Access, and available online at http://collections.plos.org/translational-bioinformatics.

ADDITIONAL REFERENCES

1. Bessant C, Oakley D, Shadforth I. Building Bioinformatics Solutions with Perl, R and SQL, 2nd ed. April 2014. ISBN-13: 978-0199658565
2. Jones NC, Pevzner PA. An Introduction to Bioinformatics Algorithms (Computational Molecular Biology). August 2004. ISBN-13: 978-0262101066
3. Durbin R, Eddy SR, Krogh A, Mitchison G. Biologicial Sequence Analysis: Probabilistic Models of Proteins and Nucleic Acids. May 1998. ISBN-13: 978-0521629713
4. Hirschhorn JN, Gajdos ZKZ. Genome-Wide Association Studies: Results from the First Few Years and Potential Implication for Clinical Medicine (Annual Review of Medicine Book 62). Kindle edition.
5. Malcolm Campbell A, Heyer LJ. Discovering Genomics, Proteomics and Bioinformatics, 2nd ed. March 2006. ISBN-9780805382198
6. Foulkes AS. Applied Statistical Genetics wirh R: For Population-based Association Studies (Use R!) April 2009. ISBN-13: 978-0387895536
7. Gondro C. Primer to Analysis of Genomic Data Using R (Use R!) May 2015. ISBN-13: 978-3319144740
8. Additional papers provided in class.

Comments: 1 is a collection of chapters describing the implications of bioinformatics at a clinical and public health level. 2 provides a very nice coverage of Perl, R and SQL motivated by bioinformatics problems and thus, is a good way to learn 3 informatics tools that are used a lot in

biomedical informatics. 3 and 4 provide in-depth presentations of mathematical and algorithmic techniques used for sequence alignment problems. They are pretty heavy from a theoretical standpoint, but are invaluable for those who are interested in in-depth coverage of modern bioinformatics. 5 is a short read discussing the implications of GWAS studies from a clinical standpoint. 6 is a basic introduction to a variety of bioinformatics issues, and is very hands on with associated web-based problems. 7 and 8 provide a deep coverage of R for bioinformatics and applications, and are a must for those interested in doing advanced data analysis of biomedical data.

COURSE DESCRIPTION: this course covers the fundamental issues of bioinformatics and how they apply to translational and clinical problems. The course is organized in 4 parts: sequence analysis, databases and ontologies, genome-wide association and linkage analysis, and networks. Each part will include coverage of the computing and mathematical concepts used, and motivated by the actual underlying bioinformatics questions.

PURPOSE OF THE COURSE: the purpose of this course is to give students a broad overview of the field of bioinformatics, and the tools commonly used in bioinformatics, as well as their applications to practical biomedical issues, diseases, population health, drug discovery, etc. The students will also be exposed to a variety of open-source programs for sequence alignment, SNPs discovery, and on how to access and analyze data from large biological databases, for translational and clinical research.

COURSE OBJECTIVES: Since the discovery of the double helix structure of DNA by Watson and Crick in 1953, the advances made in molecular biology have been gigantic. However, it became clear very soon that biologists, mathematicians, computer scientists, informaticians (but also chemists, biochemists etc.) should interact together to advance the state of the art further. The development of computational tools in the last couple of decades has been one of the main factors for the birth of bioinformatics, and the use of advanced computational tools and techniques in Biology. The objective of this course is to present the students with the fundamentals ideas of bioinformatics and how they related to translational and clinical questions. By the end of the course, students will be able to:

1. Describe and apply a broad range of bioinformatics algorithms
2. Apply informatics techniques to retrieve, store, and analyze biomedical data
3. Use open-source tools and open access databases to find and analyze data of translational and clinical importance
4. Describe the current trends and problems of bioinformatics and how they relate to clinical issues, population health and public health.

COURSE POLICIES

ATTENDANCE POLICY: Class attendance is mandatory. Excused absences follow the criteria of the UF Graduate Catalogue (e.g., illness, serious family emergency, military obligations, religious holidays), and should be communicated to the instructor prior to the missed class day when possible. UF rules require attendance during the first two course sessions. Missing more than three scheduled sessions will result in a failure. Regardless of attendance, students are responsible for all material presented in class and meeting the scheduled due dates for class assignments. Finally, students should read the assigned readings prior to the class meetings, and be prepared to discuss the material for each session.

QUIZ/EXAM POLICY: unless otherwise stipulated, all assignments are individual assignments. Students are forbidden to collaborate or consult with one another on such assignments. Students must of course follow the University Policy on Academic Misconduct, which includes but is not limited

to prohibition of plagiarism. All assignments are due at the beginning of class. All assignments have to be turned in to pass the class. NO exception.
MAKE-UP POLICY: Students are allowed to make up work only as the result of illness or other unanticipated circumstances. In the event of such emergency, documentation will be required in conformance with University policy. Work missed for any other reason will earn a grade of zero.

TERM PROJECT POLICY: the final assignment for this course is a group project, which includes a paper assignment, a programming assignment, and a presentation. As such, this is a collaborative project. Students are expected to work on this project in a professional manner, and are expected to clearly delineate roles and responsibilities.

Note: Requirements for class attendance and make-up exams, assignments, and other work in this course are consistent with university policies that can be found in the online catalog at: http://gradcatalog.ufl.edu/content.php?catoid=5&navoid=1054.

UF POLICIES

UNIVERSITY POLICY ON ACCOMMODATION STUDENTS WITH DISABILITIES: Students requesting accommodation for disabilities must first register with the Dean of Students Office (http://www.dso.ufl.edu/drc/). The Dean of Students Office will provide documentation to the student who must then provide this documentation to the instructor when requesting accommodation. You must submit this documentation prior to submitting assignments or taking the quizzes or exams. Accommodations are not retroactive, therefore, students should contact the office as soon as possible in the term for which they are seeking accommodations.

UNIVERSITY POLICY ON ACADEMIC MISCONDUCT: Academic honesty and integrity are fundamental values of the University community. Students should be sure that they understand the UF Student Honor Code at http://www.dso.ufl.edu/students.php. You are expected and required to comply with the University's academic honesty policy (University of Florida Rules 6C1-4.017 Student Affairs: Academic Honesty Guidelines, available at http://regulations.ufl.edu/chapter4/4017.pdf). Cheating, plagiarism, and other forms of academic dishonesty will not be tolerated. Note that misrepresentation of the truth for academic gain (e.g., misrepresenting your personal circumstances to get special consideration) constitutes cheating under the University of Florida Academic Honesty Guidelines

NETIQUETTE – COMMUNICATION COURTESY: All members of the class are expected to follow rules of common courtesy in all email messages, threaded discussions, and chats. The first instance of clearly rude and/or inappropriate behavior will result in a warning. The second instance will result in a deduction of five percentage points (20 points) from your overall grade. The third instance will result in a drop of a letter grade (A to B, A- to B-, and so on).

GETTING HELP

For issues with technical difficulties for E-learning in Sakai, please contact the UF Help Desk at:

- Learning-support@ufl.edu
- (352) 392-HELP - select option 2
- https://lss.at.ufl.edu/help.shtml

Any requests for make-ups due to technical issues MUST be accompanied by the ticket number received from LSS when the problem was reported to them. The ticket number will document the time and date of the problem. You MUST e-mail your instructor within 24 hours of the technical difficulty if you wish to request a make-up. Other resources are available at http://www.distance.ufl.edu/getting-help for: • Counseling and Wellness resources • Disability resources • Resources for handling student concerns and complaints • Library Help Desk support Should you have any complaints with your experience in this course please visit http://www.distance.ufl.edu/student-complaints to submit a complaint.

GRADING POLICIES

Your semester grade will be based on a combination of homework assignments, a group project, a midterm, and a final exam.

1. Project: 50%
2. Papers (3): 30%
3. Final: 20%

Notes: homework assignments will lead to class discussion. Your participation during these discussions is an effective part of the grade you receive for each assignment. Instructions for each assignment, test and for the project will be carefully described when assigned.

SCALING

Letter grade	Grade points	Grade Percentage
A	4.0	95-100
A-	3.67	90-94
B+	3.33	87-89
B	3.0	83-86
B-	2.67	80-82
C+	2.33	77-79
C	2.0	73-76
C-	1.67	70-72
D+	1.33	67-69
D	1.0	63-66
D-	0.67	60-62
F	0	<60

COURSE SCHEDULE (TENTATIVE)

Week	Topic	Notes
Wk 1	Overview of bioinformatics and principal applications: sequencing, Microarray, 'omics' fields, systems biology, data mining. Relationships to diseases and health.	Project intro. Team assignments
Wk 2	Translational Bioinformatics: Past, Present, and Future: http://www.sciencedirect.com/science/article/pii/S1672022916000401 (J. Tenebaum, Genomics Proteomics Bioinformatics 14, 2016, pp31-41)	
Wk 3	Biomedical Knowledge Integration Chap 1 of reference text P. Payne	
Wk 4-5	Data, Molecules, and Diseases Chap 2, 3, 4 of reference text C Greene et al.	
Wk 5-6	Sequencing, Assembly, Interpretation Chap 8 of reference text J. Kim	Guest Lecture Dr. T. Magoc (tentative)
Wk 7-8	Genome-Wide Association Studies Chap 11 of reference text W. Bush et al.	
Wk 9-10	Network Approaches to Diseases Chap 5 of reference text D. Cho et al.	
Wk 11	Analyses Using Disease Ontologies Chap 9 of reference text N. Shah et al.	
Wk 12	EHRs and 'omics' fields Chap 13 of reference text J. Denny	
Wk 13	Human Microbiome Analysis Chap 12 of reference text X. Morgan et al.	Guest Lecture Dr. D. Lemas (tentative)
Wk 14	Cancer and Translational Bioinformatics Chap 14 of reference text M. Vazquez et al.	
Wk 15	Project Presentations	