Systems Medicine

Assignment II and Organization

Select one of the provided papers and discuss it following the layout of "critical evaluation of a paper" (see class material). Provide a written reflection of your work via a comprehensive slides deck, in which you have woven in the guiding questions/themes that match the paper of your choice. These slides will contribute to your "written exam".

Form groups of 2 students. For each group: compile a group presentation that is composed of the following. A) A portion of each individual's slides, addressing 3 series of questions in depth (3 by person 1 and 3 by person 2); B) Slides that form the connection between the two papers. Pool the slides you will present (student 1, student 2, joint), name it "Bioinformatics Applications Assignment II _two names" and submit it to Prof Van Steen (kristel.vansteen@uliege.be) with the subject title "Bioinformatics Applications Assignment II", by Sunday night April 18. During the class of April 21 each student in a team of 2 is expected to present his/her own part; the common part is presented jointly. For each speaker, the entire presentation takes about 15-20min. Thereafter, these presentations will be taken as a starting point for Q&A and a discussion to clarify unknown concepts, while making links to the course materials and the latest developments in the field.

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Papers

Example case studies:

https://erj.ersjournals.com/content/52/suppl_62/PA1278
https://www.ncbi.nlm.nih.gov/pubmed/27901618

An adapted list will be provided after class 1 – based on individual interests

(see http://bio3.giga.ulg.ac.be/index.php/education/courses-at-ulg/)

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10 series of guiding questions

(Adapt the questions to the "systems medicine" concept covered by your paper. Each question starts with the main theme of the question, which is relevant to different aspects of systems medicine.)

1. Setting: Describe the biological question(s) and the set-up of the study. Highlight specific differences with a classical genome-wide association study where the aim is to find genetic predispositions to disease.
2. Definition: Give an epidemiology oriented definition of "interactions/epistasis". Is it different from effect modification? Give and discuss different definitions of "interactions/epistasis", in different contexts. Is there a difference between statistical and genetic epistasis? Is it easy to translate statistical epistasis into biological epistasis? How can this translation be facilitated?
3. Adding levels of complexity: In what ways will a gene-environment interaction study be different (more complex? less complex?) than a gene-gene interaction study?
4. Computational efficiency: What is meant by an exhaustive search? Is this feasible in the context of a genome-wide setting?
5. Networks: Does it make sense to investigate higher-order interactions?

6. Simplistic versus more elaborate, sophisticated methods: What are the criticisms to traditional regression-based approaches in the context of genome-wide interaction studies (GxG or GxE) and can you give alternative methods to deal with the abundance of complex data patterns?
7. One popular method/approach singled out: What is multifactor dimensionality reduction? What are its advantages and limitations? What are some advantages and limitations of machine learning / deep learning methods?
8. Replication: Replication and validation are important components of any genetic association study. What would replication of a genome-wide interaction study involve?
9. Experimental validation: What is the state of the art? What are the problems when trying to experimentally validate findings from big Omics (integrative) studies?
10. Personalized medicine: Can you highlight the differences between “genomics for personalized medicine” and “public health genomics”?