

Clinical and Research Genomics Assignment #6

From Lecture_16-18 (April 26th): Personalized Medicine and Disease Classification

Assignment: Use Omicia software to identify and interpret disease-causing variants
Due Date: 5:00PM on May 8th

Case study: female patient presenting with pruritus and failure to thrive. In your project, you have access to variant reports from exome sequencing data for the patient and both parents, who are unaffected. Your assignment is to diagnose the patient and identify any genetic variants that explain her symptoms.

1. Create an Omicia account and log in to Opal Clinical. If you do not receive an invitation to join Omicia by May 1st, please email prv2004@med.cornell.edu.
2. Explore tools and apps available (VAAST solo, VAAST trio, Clinical report generator, Phevor phenotype analysis). In addition to the trio for the case study, there is also a breast cancer sample with targeted sequencing in your project for further exploration.
3. Run these analyses on the trio to identify the causative variant(s), and generate a report that includes an explanation of your pipeline, biological significance of variants identified, and supporting evidence such as coverage and quality of variant base calls and Manhattan plots of gene-symptom associations.

Please hand the assignment on the day of the lecture, or email if you cannot attend. For any questions, please contact Priyanka Vijay (prv2004@med.cornell.edu), Ebrahim Afshinnekoo (eba2001@med.cornell.edu), Alexa McIntyre (abm237@cornell.edu), or Professor Mason (chm2042@med.cornell.edu).