GENETICS, BIOINFORMATICS & SYSTEMS BIOLOGY COLLOQUIUM

THURSDAY, February 25
12:00-1:00 PM
Held on Zoom

Click Here for Zoom Link!
Meeting Password: IGM

MANUEL RIVAS, PhD
guest speaker
Assistant Professor
Department of Biomedical Data Science
Stanford University

Talk Title: Genomic prediction and inference from population-scale datasets

Clinical laboratory tests are a critical component of the continuum of care and provide a means for rapid diagnosis and monitoring of chronic disease. In this study, we systematically evaluated the genetic basis of 35 blood and urine laboratory tests measured in 358,072 participants in the UK Biobank and identified 1,857 independent loci associated with at least one laboratory test, including 488 large-effect protein truncating, missense, and copy-number variants. We then causally linked the biomarkers to medically relevant phenotypes through genetic correlation and Mendelian Randomization. Finally, we developed polygenic risk scores (PRS) for each biomarker and built multi-PRS models using all 35 PRSs simultaneously. We assessed sex-specific genetic effects and find striking patterns for testosterone with marked improvements in prediction when training a sex-specific model. We found substantially improved prediction of incidence in FinnGen (n=135,500) with the multi-PRS relative to single-disease PRSs for renal failure, myocardial infarction, type 2 diabetes, gout, and alcoholic cirrhosis. Together, our results show the genetic basis of these biomarkers, which tissues contribute to the biomarker function, the causal influences of the biomarkers, and how we can use this to predict disease.

Faculty Host: Amit R. Majithia, MD
Assistant Professor, Dept of Medicine

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Organization Committee: J. Gleeson, F. Furnari, A. Majithia, T. Gaasterland
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