Stony Brook University

Biomedical Informatics Special Seminar

Vikas Pejaver, Ph.D.

Postdoctoral Scholar, Department of Biomedical Informatics and Medical Education, University of Washington

Machine Learning on Secondary Data for Rare Genetic Diseases

Friday, April 17, 2020 3 pm—4 pm

Abstract:

Rare genetic diseases are estimated to collectively affect 6-8% of the global population, placing a substantial burden on the healthcare system and patients themselves. Despite advances in high-throughput sequencing and the systematization of knowledge, our understanding of, and thus our ability to diagnose the 7,000-8,000 known rare diseases needs to be greatly improved. A key challenge is the limited data available for the use of traditional approaches towards the study of such disorders. More specifically, while data on disease-associated genetic variants are accumulating, the molecular mechanisms that link such variants to observable disease phenotypes is limited. In this talk, I will discuss how computational methods that leverage retrospectively collected secondary data, ranging from accumulated literature on genetic variants and protein function to phenotypic information in patient health records in medical systems' enterprise data warehouses, can help overcome this challenge. I will also briefly discuss how machine learning approaches need to be adapted for situations where one has little information on the data generating process. While such situations arise in several areas of the biomedical and health sciences, I will highlight the potential role of machine learning in the discovery, diagnosis and treatment of rare genetic diseases.

Bio:

Dr. Vikas Pejaver is a postdoctoral scholar at the Department of Biomedical Informatics and Medical Education and the eScience Institute at the University of Washington. Under the mentorship of Dr. Sean Mooney, he is working on the development and application of machine learning methods for genomic, molecular and health record data to accelerate the discovery, diagnosis and treatment of rare genetic diseases. He is a recipient of a Moore/Sloan and Washington Research Foundation Innovation in Data Science Postdoctoral Fellowship and a K99/R00 Pathway to Independence Award from the National Library of Medicine at the National Institutes of Health. Dr. Pejaver received his B.E. in Biotechnology with Distinction from the People's Education Society (PES) University in Bengaluru, India, and M.S. in Bioinformatics at Indiana University, Bloomington. At Indiana, he went on to complete his Ph.D. in Informatics in Dr. Predrag Radivojac's research group.

Remote Access

Join Zoom Meeting https://zoom.us/j/382860134?pwd=WGRoWTFBcDR0QkpDbkYwTHJxNUxGdz09 Meeting ID: 382 860 134

One tap mobile +16465588656,382860134# US (New York)

Questions? Please call the Biomedical Informatics Department at 631-638-2590.