COLUMBIA
CENTER FOR PRECISION MEDICINE AND GENOMICS
IN THE DEPARTMENT OF MEDICINE

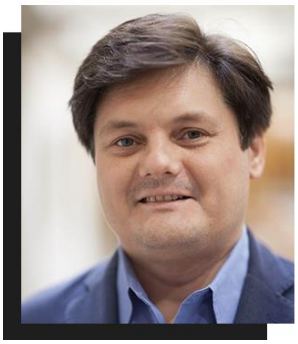
COLUMBIA
IRVING INSTITUTE FOR CLINICAL AND TRANSLATIONAL RESEARCH

COLUMBIA
PRECISION MEDICINE

Seminars in Precision Medicine

Sponsored by the Center for Precision Medicine and Genomics, the Precision Medicine Resource of the Irving Institute for Clinical and Translational Research, and the Columbia Precision Medicine Initiative

"Bridging Statistical Genetics and Biology using Rare Variants and De Novo Mutations"



Shamil Sunyaev, PhD

Professor of Biomedical Informatics, Harvard Medical School Professor of Medicine, Brigham and Women's Hospital

Biological knowledge is commonly organized in the form of networks or pathways. New network models are continuously generated in model organism and cell system screens including Perturb Seq experiments. To establish relevance of network and pathway models to human phenotypes, we developed a rare variant association test that takes into account network connectivity. We applied the test to blood lipid traits, coronary heart disease, myocardial infarction and breast cancer phenotypes. Applications to Parkinson disease motivated follow up experimental work showcasing an iterative analysis involving statistical genetics testing and gene editing experiments. We also developed new, well-calibrated statistical methods for prioritizing rare disease genes with de novo recurrence and compound heterozygosity. A new method detects pathways enriched with candidate and known diagnostic genes. These statistical tests use a biologically informed model of human mutation rate along the genome (Roulette).

Date: Thursday, December 12th, 2024

Time: 4:00 – 5:00 p.m.

Location: Presbyterian Hospital, PH20-200

Registration Link: https://events.columbia.edu/go/SSunyaev

Contact Information:

Anna Abreu

Aa5133@cumc.columbia.edu

Scan for more information: