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Virtual BSC RS/Life Session/BioInfo4Women Seminar: Functional variation in the human genome: lessons from the transcriptome

Objectives

Abstract: Detailed characterization of molecular and cellular effects of genetic variants is essential for understanding biological processes that underlie genetic associations to disease. A particularly scalable approach has been linking genetic variants to effects in the transcriptome that is amenable for scalable measurements in human populations and in experimental settings, including at the single cell level. Our multi-omic analysis in human cohorts in the TOPMed project has identified genetic and environmental effects on molecular variation together with their complex interplay with clinical phenotypes. Furthermore, in this talk I will discuss how CRISPRi silencing of regulatory elements followed by single-cell analysis provides novel insights of mechanisms of genetic associations to complex traits. Altogether, these diverse approaches for integration genome and transcriptome data uncover functional genetic architecture of human traits, and enhances our understanding of both basic biology and precision medicine applications.



Short bio: Tuuli Lappalainen, PhD, is

a Professor at KTH Royal Institute of Technology, the Director of the Genomics Platform and the National Genomics Infrastructure of SciLifeLab, Sweden, and an Associate Faculty Member at the New York Genome Center. Dr. Lappalainen received her PhD in Genetics from the University of Helsinki, followed by postdoctoral research at the University of Geneva and Stanford University. Her research focuses on functional genetic variation in human populations and its contribution to human traits and diseases, which her lab studies using both computational and experimental approaches. She has pioneered in integrating large-scale genome and transcriptome sequencing data to understand how genetic variation affects gene expression, which gives insight to biological mechanisms underlying genetic disease risk. She has contributed to many of the most important international research consortia in human genetics, including the 1000 Genomes Project, the Geuvadis Consortium, the GTEx Project, MoTrPAC, and TOPMed. She is a principal investigator of numerous NIH grants and a recipient of the Leena Peltonen Prize for excellence in human genetics, and the Harold and Golden Lamport Award in Excellence in Basic Research.

Speakers

Speaker: Tuuli Lappalainen, Professor, KTH Royal Institute of Technology. Director, National Genomics Infrastructure and Genomics Platform, SciLifeLab. Associate Faculty Member, New York Genome Center
Host: Marta Melé, Transcriptomics and Functional Genomics Lab Group Leader
Barcelona Supercomputing Center - Centro Nacional de Supercomputación

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