SORS: Functional variation in the human genome: lessons from the transcriptome

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, which is amenable for scalable measurements in human populations and in model systems, including at the single cell level. Here, I will describe recent advances in our long-term work to characterize genetic associations to the transcriptome and other molecular traits, as well as our recent work on CRISPR-based perturbation of gene expression levels in cellular models. Altogether, integrating insights from these diverse approaches uncovers functional genetic architecture of human traits and the molecular and cellular mechanisms that mediate these effects.

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 a Senior Associate Faculty Member at the New York Genome Center. Dr. Lappalainen received her PhD in Genetics from the University of Helsinki. Her research focuses on functional genetic variation in human populations and its contribution to human traits and diseases. She has contributed to many international research consortia in human genetics, including the 1000 Genomes Project, the Geuvadis Consortium, and the GTEx Project. She has received multiple prizes for her contributions to the field.

Speakers

Speaker: Tuuli Lappalainen, PhD. Professor at KTH Royal Institute of Technology
Host: Marta Melé. Leading Researcher, Transcriptomics and Functional Genomics Lab. Life Sciences Department, BSC