

# The Leading Strand Educational Webinar Series



**Tuuli Lappalainen, PhD**

Professor, KTH Royal Institute of Technology  
Director, National Genomics Infrastructure & Genomics Platform, SciLifeLab  
Associate Faculty Member, New York Genome Center  
New York, USA

## *Functional variation in the human genome: lessons from the transcriptome*

12PM, EST  
May 27, 2022  
Host: Dr. Monica Justice

Register in advance for this webinar: [https://us06web.zoom.us/webinar/register/WN\\_vXOheKQmTnOAG2Mt1BZbJw](https://us06web.zoom.us/webinar/register/WN_vXOheKQmTnOAG2Mt1BZbJw)

*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.*