



# Genome wide studies of promoter and enhancer activities: clinical applications

(

Cellular development and tissue homeostasis requires coordinated expression of genes, facilitated by regulation of promoter and enhancer activity. Genetic and epigenetic perturbation of this regulation may cause aberrant gene expression and lead to impaired cell formation and function. This seminar will introduce ways in which promoter and enhancer expression profiling at bulk and single cell level together with bioinformatics analysis can help formulating powerful hypotheses about disease. I propose technologies and suggest workflows for different use cases aiming to identify the regulatory networks behind cellular disease states. This includes inferring key transcriptional regulators and non-coding RNAs modulating or modulated by disease conditions, and assigning function to genetic variants associated to disease traits.