



CPH Seminar in Precision Medicine

“Exploring Disease Mechanisms using Genetically Regulated Endophenotypes”

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We have experienced an unprecedented progress in our ability to understand the function of the genome. Advances in high throughput technology have enabled us to identify thousands of genomic loci indisputably associated with a broad spectrum of human complex traits. However, the translation of this knowledge has been slow because of the gaps in our understanding of the mechanism that link genome variation with phenome variation. There is general consensus that gene expression regulation is an important component of this mechanism. To test the mediating role of gene expression traits we have pioneered the use of genetically predicted transcriptome, which is becoming part of standard post GWAS analysis. In this talk, I will describe the basics and motivation behind our method PrediXcan, present extensions of the method, and discuss next steps to address some of the limitations of the method.

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