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Functional variation in the human genome: lessons from the transcriptome

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Detailed characterization of cellular effects of genetic variants is essential for understanding biological processes that underlie genetic associations to disease, to improve the interpretation of the personal genome, and to characterize the genetic architecture of molecular variation. This has inspired large consortium projects to create and integrate population-scale genome data with transcriptome data – as well as other molecular phenotype data – in human populations. The catalogs of genetic effects on the transcriptome across multiple human tissues and conditions now allows downstream discovery in diverse questions in genetics. In this talk, I will discuss the GTEx consortium efforts in cataloguing regulatory variants in the genome, and our work in mapping their role as modifiers of coding variant penetrance and as causal factors in rare disease.