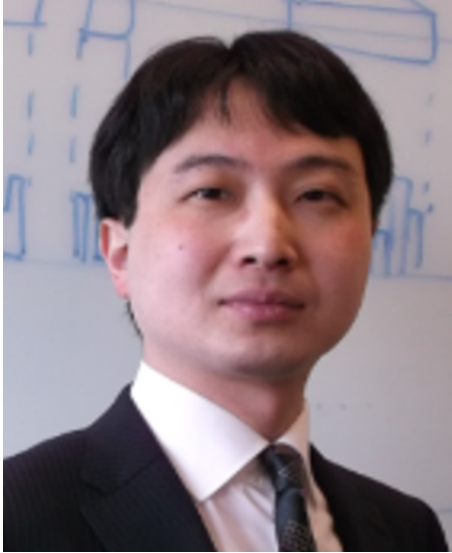


## **A new genomic and computational approach to study human genomic enhancers and its association with diseases**



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The complexity of an organism is largely determined by the variety of gene regulatory mechanisms. In particular, enhancers are key distal non-coding *cis*-regulatory elements, and enhancer dysfunction can lead to a multitude of human diseases. Here we developed native elongating transcripts-cap analysis of gene expression (NET-CAGE), a robust method to profile genome-wide 5' ends of nascent RNAs in diverse cells and tissues (Hirabayashi et al. Nature Genetics, 2019). Using NET-CAGE, we detected unstable non-coding RNAs that are derived from active enhancers with an unprecedented sensitivity, allowing us to characterize a large number of novel human enhancers. Strikingly, these enhancer regions are significantly enriched for human disease-associated genetic variants. We combine our original genomic and computational approaches to study the functionality of gene regulatory network in human health and disease.