



CPH Seminar in Precision Medicine

“Identification of disease genes for rare and undiagnosed disorders through an integrative multi-omics approach”

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Although rare diseases, by definition prescribes to uncommon pathophysiological phenotypes seen a small fraction of the population, the combined number for all rare disease groups as a whole represents a big medical challenge. It is estimated 25-30 million people in the United States are affected by rare disorders, 80% of which are thought to have a genetic origin. This highlights the need of a precise and accurate genetic diagnosis, a critical and fundamental step precedent effective clinical management or treatment.

Coupled with RNA-sequencing, we were able to interrogate non-coding variants that are previously refractory to prediction of their biological functions. An in-house pipeline was developed using Cloud-computing infrastructure to bring forth an integrative analysis utilizing both DNA-seq and RNA-seq to identify abnormal patterns associated with non-coding variants lie within potential splicing regulatory regions. Overall, we have been able to achieve a higher rate of identifying disease causing or strong candidate variants or genes than reported rate before.

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