UTHealth
The University of Texas Health Science Center at Houston
School of Biomedical Informatics

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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