Stony Brook University
The State University of New York

Department of Biomedical Informatics
School of Medicine and College of Engineering and Applied Sciences

SPECIAL SEMINAR

Simon Chong Chu, PhD
Principal Data Scientist, ROME Therapeutics

Contribution of genomic repeats to cancer and birth defects

Wednesday, May 22, 2024
2:00 pm – 3:00 pm

Join Zoom Meeting
https://stonybrook.zoom.us/j/99757826947?pwd=M2U1cjErQzZ6c0lZSkxaUjlYM1VtUT09

Meeting ID: 997 5782 6947
Passcode: 507956

Abstract: Over 50% of the human genome consists of genomic repeats, encompassing transposable elements (TEs), satellite, and short tandem repeats. Numerous diseases have been reported to be associated with genomic repeats. To characterize TE mobility, we have developed xTea, an exceptionally comprehensive TE insertion identification method that can be applied to on multi-omics data. We applied xTea on 3,244 trios from 12 birth defect and childhood cancer cohorts. We identified 162 de novo retroelements, including several potential causal ones. We observe a high de novo SVA insertion burden in both high-intolerance loss-of-function genes and exons.

TEs can also co-opt nearby genes, forming exon-trappings that alter the hosting genes. We depicted the landscape of SINE-VNTR-Alu (SVA) copies in the human genome and developed the computational method TrapHunter to identify SVA formed exon-trapping events from RNA-seq data. Applying TrapHunter to >25,000 TCGA and GTEx RNA-seq samples, we identified numerous tumor- and tissue-specific SVA exon-trapping events.

Bio: Simon currently serves as a Principal Data Scientist at ROME Therapeutics, a pioneering biotech firm focused on harnessing the potential of transposable elements in combating autoimmune diseases and cancer. Prior to his role at ROME, Simon completed his postdoctoral training at the Department of Biomedical Informatics at Harvard Medical School. During this time, he dedicated his efforts to developing novel computational methods to characterize the roles of transposable elements in cancer etiology, autism, and birth defects. Simon earned his Ph.D. from the Department of Computer Science and Engineering at UCONN, where his research centered on de novo repeats assembly and population genomics.