GENETICS, BIOINFORMATICS, & SYSTEMS BIOLOGY COLLOQUIUM

THURSDAY DECEMBER 9TH
12:00PM PST
ON ZOOM!

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PASSWORD: GENETICS

PRESENTED BY: IGM Institute for Genomic Medicine | UC San Diego Bioinformatics & Systems Biology

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PO-RU LOH, PHD

DIVISION OF GENETICS, BRIGHAM AND WOMEN'S HOSPITAL

BROAD INSTITUTE OF THE MIT AND HARVARD MEDICAL SCHOOL

PROTEIN-ALTERING VARIANTS WITH LARGE PHENOTYPIC EFFECTS: RARE SNPS, COMMON VNTRS, AND RARE CNVS

Genome-wide association studies (GWAS) have identified hundreds of thousands of associations between genetic variants and human phenotypes. However, most such statistical associations involve noncoding variants with no clear function that typically reflect correlation rather than causation. Only a small subset of associations discovered to date involve genetic variants that directly alter protein-coding sequence. We have recently undertaken several projects to expand this set of more-interpretable genetic associations by analyzing variation in parts of the coding genome previously inaccessible to extremely large-scale (N~500K) analysis: namely, very rare coding variants and two types of structural variants. These analyses, made possible by new computational methods that leverage haplotype-sharing within biobank cohorts, have explained some of the strongest common-variant associations with human traits and identified many rare coding variants with large effect sizes (>0.5 standard deviations). These results likely represent just the beginning of the insights that will be uncovered as genetic association studies expand to truly consider genome-wide variation.

Organization Committee: J. Gleeson, J. Sebat
GBSBC Seminar Coordinator: R. White

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