Speaker: Po-Ru Loh, Harvard Medical School

Date: Wednesday, March 2, 2022 **Time:** 11:30 AM to 1:00 PM

Host: Bonnie Berger

Title: Haplotype-informed discovery of hidden genetic variants influencing human traits

Abstract: Genetic association studies have discovered hundreds of thousands of common single-nucleotide polymorphisms (SNPs) associated with human phenotypes. However, such associations have generally been difficult to interpret, often only providing hints of other nearby genetic variation that causally modifies traits. In this talk, I will describe progress on ascertaining and evaluating the effects of three understudied types of genetic variants: inherited copy-number variants (CNVs), noncoding variable number tandem repeats (VNTRs), and rare coding SNPs. This work has been powered by statistical methods that leverage haplotype-sharing among distantly related individuals in large cohorts.