

Mosaicism in genetics refers to the state in which cells of a single individual belong to two or more sub-populations with distinct genotypes. Mosaicism can arise either through mutations early in development, or through mutations later in life that subsequently undergo clonal expansion (e.g., cancer). In this talk, I will present new statistical methods that leverage population-based haplotype phasing to enable detection of mosaic chromosomal alterations present at very low cell fractions. I will describe insights into the causes and consequences of age-related clonal hematopoiesis revealed by this approach and discuss applications to mosaicism in other tissues.