





Homozygosity disequilibrium (HD), defined by a non-random pattern of sizable run of homozygosity in the genome, plays important roles in medical genomics and population genomics. The major genetic mechanisms of HD include but not limited to autozygosity, natural selection, and chromosomal aberrations. We developed statistical methods and software to dissect the whole-genome patterns of HD. Performance of the tools has been carefully evaluated by simulation studies and real data analyses of whole-genome single nucleotide polymorphism and next-generation sequencing data. In this talk, some examples of achievements will be presented: We identified genomic segments bearing HD, found familial aggregation of HD, derived the genomic distribution of HD, uncovered disease-associated regions of HD, detected samples with structural alterations and/or unusual genotypic patterns, classified samples with close structure of HD, and explained roles of HD in gene expression regulation and pharmacoepigenomics.