Deciphering Population Genomics and Medical Genomics through Homozygosity Disequilibrium Using Whole-Genome Single Nucleotide Polymorphism and Next-Generation Sequencing Data

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Abstract

Homozygosity disequilibrium (HD), defined by a sizable tract of homozygosity deviated from a random distribution in the genome, plays important roles in population genomics and medical genomics. We developed LOHAS software to dissect the whole-genome patterns of HD (http://www.stat.sinica.edu.tw/hsinchou/genetics/loh/LOHAS.htm). LOHAS is applied to analyze whole-genome single nucleotide polymorphism and next-generation sequencing data in studies of human populations, acute lymphoblastic leukemia, rheumatoid arthritis, and hypertension. In these applications, we detect genomic segments bearing HD, uncover their disease association, identify samples with structural alterations and/or unusual genotypic patterns, cluster samples with close structure of HD, and find heredity of HD successfully.