Copy Number Variation Detection with Estimated Parameters Using Robust Distribution

High-density single nucleotide polymorphism arrays (SNP) are effective in identifying copy number variations (CNVs) in the whole genome. Existing methods to detect CNVs primarily use log R ratio and B allele frequency while sample-specific parameters are predominantly excluded. In this paper, we propose a robust hidden Markov model in which parameter values are estimated using a computationally efficient EM algorithm. Our model also takes into account inter-marker correlation. Finally, we demonstrated accuracy of our method in detecting various CNV regions using simulated data.