An integrated approach for the assessment of chromosomal abnormalities

Many chromosomal abnormalities such as amplifications, deletions, and copy-neutral loss of heterozygosity have been associated with disease. High-throughput single nucleotide polymorphism (SNP) arrays are useful for the genome-wide assessment of such abberations. We present some integrated approaches for the assessment of such chromosomal abnormalities based on SNP chip estimates of genotype, copy number, and uncertainty measurements. We will also discuss how to assess parent-of-origin effects in trio data.