Microarray-based maps of copy-number variant regions in European and sub-saharan populations

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The genetic basis of phenotypic variation can be partially explained by the presence of copy-number variations (CNVs). Currently available methods for CNV assessment include high-density single-nucleotide polymorphism (SNP) microarrays that have become an indispensable tool in genome-wide association studies (GWAS). However, insufficient concordance rates between different CNV assessment methods call for cautious interpretation of results from CNV-based genetic association studies. Here we provide a cross-population, microarray-based map of copy-number variant regions (CNVRs) to enable reliable interpretation of CNV association findings. We used the Affymetrix Genome-Wide Human SNP Array 6.0 to scan the genomes of 1167 individuals from two ethnically distinct populations (Europe, N = 717; Rwanda, N = 450). Three different CNV-finding algorithms were tested and compared for sensitivity, specificity, and feasibility. Two algorithms were subsequently used to construct CNVR maps, which were also validated by processing subsamples with additional microarray platforms (Illumina 1M-Duo BeadChip, Nimblegen 385K aCGH array) and by comparing our data with publicly available information. Both algorithms detected a total of 42669 CNVs, 74% of which clustered in 385 CNVRs of a cross-population map. These CNVRs overlap with 862 annotated genes and account for approximately 3.3% of the haploid human genome.We created comprehensive cross-populational CNVR-maps. They represent an extendable framework that can leverage the detection of common CNVs and additionally assist in interpreting CNV-based association studies.
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