

P047 Global genomic variation in copy number
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Copy number variation (CNV) within the human genome is of functional importance and yet remains grossly under-ascertained. To redress this imbalance and generate a CNV map of the genome, we screened all 270 individuals from the four HapMap populations with ancestry in Europe, Africa and East Asia for copy number variation using two complementary technologies: the Affymetrix 500k SNP genotyping platform and comparative genome hybridisation on a spotted microarray containing ~27,000 clones representing the genome tilepath. We rigorously assessed the rate of false positive CNV calls by several independent means, including the validation of over 100 loci by quantitative PCR and FISH. We identified over a thousand CNVs in these four populations, many of which have not previously been identified, and which contain hundreds of genes and non-coding functional sequences. We ascertained the impact of these CNVs on the reliability of SNP genotypes from the International HapMap Project. We also delineated the linkage disequilibrium properties of hundreds of these CNVs, and identified tagging SNPs. Finally, we identified the genomic regions that show the most dramatic variation in copy number between populations, and explored selective explanations for these observations. Our data is released in public genome browsers for the community. This is the first global, genome-wide assessment of CNV and its relationship to haplotypic structure, and provides an important resource for the understanding of how genome structure impacts on biological function.