MULTIPLEXED PRIMER EXTENSION ASSAYS FOR GENOTYPING OF Y-CHROMOSOME AND AUTOSOMAL SNPs

David Carlson, Christopher Rowe and Zoe D'Aoust
Marligen Biosciences, Ijamsville, MD

Suspension arrays have been developed for rapid genotyping of SNP loci. Commercial applications of this technology have been built on allele-specific oligonucleotide hybridization. We have developed next generation suspension array assays based on allele-specific primer extension (ASPE). The new assays take advantage of the specificity of DNA polymerases to increase signal-to-noise ratios. They also allow utilization of brighter fluorescent labels that boost sensitivity. We have utilized multiplexed ASPE reactions on suspension arrays to rapidly screen Y-SNPs, which provided a rapid identification of the racial derivation of the paternal lineage. The frequencies of the Y-haplogroups in Caucasian and African American samples are consistent with commonly accepted estimates of admixture in the U.S. We have also applied the ASPE chemistry to developing multiplexed assays for autosomal SNPs that will be useful for general identification purposes. The assays cleanly discriminated heterozygotes and homozygotes. We have identified a panel of 60 SNPs that each have a frequency of 40% to 50% in the three major U.S. population groups. Taken together, this set of SNPs is a powerful tool for high-throughput human identification.