

hYperplex – A Y-STR MULTIPLEX GENOTYPING PANEL WITH HYPER-DISCRIMINATORY CAPACITY

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Introduction: Y chromosome Short Tandem Repeat (Y-STR) testing has been broadly exploited in forensic casework for profiling male DNA especially in the presence of excessive amounts of female DNA or mixtures of more than one male biological sample as in some sexual assault cases. The discriminatory capacity of commercially available Y-STR kits has some limitations due to the moderate diversity values of loci used in the kit. Development of a new Y-STR genotyping panel with high discriminatory power will provide a powerful tool for forensic case work and patrilineal ancestry analysis.

Methods: 13 extensively evaluated noncore Y-STR loci were selected and incorporated within the hYperplex panel developed at Sorenson Genomics. Primers were carefully designed based on ISFG nomenclature guidelines, re-sequencing verification and specific data quality criteria. An allelic ladder was also developed to standardize the allele calls at each locus. The study included 595 unrelated male samples derived from varying populations (Caucasian n = 134, African American n = 116, Asian n = 72, Hispanic n = 18, Mixture n = 63, Unspecified n = 192), genotyping on hYperplex, commercial Y-filer® and another set of four Y-STR multiplex panels which included 43 core and noncore loci (Y-43). The discriminatory power of hYperplex was calculated against those of Yfiler® and the in-house Y-43, Y-STR multiplexes. The allele frequencies were calculated based on the results of the 595 samples. The optimized multiplex performance studies including specificity, sensitivity and mixtures were evaluated.

Result: The hYperplex panel possesses a very higher discriminatory capacity (DC) of 99.7%, compared to the commercially available Y-filer® kit, with a DC of 96.1%, and an in-house Y-43, Y-STR multiplex test with a DC of 98.8%. The allelic ladder developed for the hYperplex test also allows for fast and confident genotyping. The assay was capable of generating a full Y-STR profile from only 66 picograms of male DNA. Male/female mixture studies indicated that full male profiles were obtained when 4ng male DNA were co-amplified in the presence of female DNA at concentrations of 8, 20, 40, 400, 2000, 4000 and 10000 nanograms respectively. Male/male mixture studies demonstrated that a full Y haplotype from a minor male component was obtained when it comprised of ½ and ¼ of total 4 ng mixed male DNA.

Conclusion: This study demonstrated that the hYperplex Y-STR system serves as a powerful tool for forensic casework as a stand-alone test or as a confirmatory method for other currently available Y-STR test kits.