CROSS VALIDATION OF ALLELE CALLING SOFTWARE FOR STR ANALYSIS IN FELON DATA BASING APPLICATIONS "ALLELECALLER" VERSUS GENOTYPER™

B.E. Ward, B. Duceman, A. Fraser, R. Brown, P. Wistort, and M. Hulick

Myriad Genetics Laboratories, Salt Lake City and New York State Police Forensic Investigation Center



"Allelecaller" is a proprietary software program that was developed specifically to produce precise, accurate allele calls in a high throughput, genotyping facility. This software application was originally utilized in a research environment to assist in human gene discovery. Typically, comprehensive genome wide searches were performed utilizing various combinations of 950 different validated STR markers. Currently, Allelecaller is used in a facility where 2.5 million human genotypes are determined annually.

Allelecaller was also designed to provide enhancements to existing software programs such as PE Applied Biosystems Genotyper™. Two aspects of improved functionality of Allelecaller over commercially available software include enhancements to data extraction and unambiguous identification of authentic fragments from which to make allele calls.

Allelecaller is currently being utilized for STR analysis in felon data basing applications. Specifically, samples are prepared using commercially available kits (PE Biosystems) and evaluated for all 13-core STR loci on a highly automated analysis platform. This platform incorporates Tecan robots, MJ tetrad thermocyclers, and ABI 377 Automatic DNA sequencers. All of these components interact with a comprehensive laboratory information management system. Data generated on the automatic sequencers is transferred into Allelecaller software for generation of final allele calls for felon data base applications.

One final stage in evaluation of commercial platforms and Allelecaller software for applications to STR analysis of the 13 core loci for convicted felon database application was a three way–cross validation. Eighty-eight samples were analyzed independently at two different facilities for all 13 core loci plus amelogenin. Allele calls were determined blindly by Genotyper® at both facilities and at Allelecaller at one facility (MGL). Results demonstrated complete concordance for all allele calls at every loci. This level of concordance indicates that proficiency between the analysis systems is greater than 99.86% (95% confidence).