FORENSIC SOFTWARE – SNPS IN FLANKING REGION ENHANCES DISCRIMINATION POWER

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Single nucleotide polymorphism (SNPs) are the most prevalent genetic markers in genome that are usually bi-allelic with varying allele frequencies for different populations. SNPs linked to the flanking regions of STR markers can be leveraged to improve the discrimination power of the same marker and for mixture deconvolution in forensic casework.
Massively parallel sequencing (MPS) allows concurrent analysis of genetic markers relevant for human identification. MPS technology has the ability to capture flanking regions variations of STR markers in addition to the STR repeat sequence.

A total of 55 variations were identified in flanking regions that includes 54 SNPs and 1 deletion marker. 8 SNPs have global alternate allele frequency above 10%. Out of 36 STR markers, 22 of them have SNPs in flanking regions which is 61% of markers. STR & SNP markers were sequenced using forensically relevant Precision ID panels combined with the Ion Chef™ System and Ion S5 sequencer System and the data analysis was done using Converge ™. Converge ™ application displays SNPs in flanking regions for each STR allele, if available, along with coverage & quality of each SNP detected. A graphical representation of SNP in flanking regions is also displayed that makes analysis easy.

Variations detected in flanking regions increased the number of STR alleles of the same size thereby increasing the heterozygosity of the STR marker. Single source sample can have isometric heterozygotes for the same STR allele with respect to the SNP detected. These variations can clearly indicate the contribution of a forward or reverse stutter in a true allele that cannot be deciphered with just the STR repeat number and the sequence. SNPs are a good tool for separating minor and major contributor in case of a shared STR allele as well as to distinguish an allele of a minor contributor and the stutter of a major contributor. Analysis of mixture sample with major and minor contributors in ratio 1:10 & 1:20 indicates the ability to detect minor contributor alleles using STR-SNP alleles. Single source samples analysis shows SNP calling accuracy of 97.6%.

Genotyping these variants has a great potential in unravelling the number of contributors and assistance in mixture deconvolution.

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