AN OPEN FRAMEWORK APPLICATION FOR ANALYZING STRs AND OTHER FORENSIC MARKERS FROM SECOND GENERATION SEQUENCING DATA

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Several recent studies have demonstrated the ability to type forensic identity markers from second generation sequencing (SGS) data. However the methods for typing various marker classes have been implemented differently in various software packages, causing the forensic analyst to potentially switch between disparate software packages with significantly different user interfaces. We present here a refinement of a previously published method for determining STR allele repeat patterns from SGS data and demonstrate within the context of an open application framework designed to provide a single and consistent user experience while analyzing any forensic endpoint. Software programs tailored for specific forensic analyses are implemented within this genomics workbench application as integrated plug in modules. In this study, we present SGS data analysis of forensic markers using the STR identification module, a genomic-compatible data repository, and a genomic-compatible data exchange format. The performance characteristics of the STR analysis module are described including handling of sequence variants, and observed signal to noise. The forensically-oriented genomics format for data exchange is a direct extension of, and is backwards compatible with, the current CMF 3.2 and CMF 4.1 specifications for exchanging STR and mitochondria typing data. In addition, this format provides the specification for exchanging STR and SNP allele sequence information. This open framework architecture with analysis and data management programs designed for forensically-oriented SGS data offers a comprehensive solution in next generation analysis of forensics data to be implemented by both forensic DNA testing facilities and forensic research laboratories.