IMPLEMENTATION AND VALIDATION OF AN AUTOSOMAL AND Y SHORT TANDEM REPEAT WORKFLOW FOR MASSIVELY PARALLEL SEQUENCING

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While short tandem repeat (STR) typing by capillary electrophoresis (CE) remains the primary method for DNA typing in forensic genomics, the amount of information is limiting. STR typing by massively parallel sequencing (MPS), by contrast, yields both size- and sequence-based information, allows for the processing of larger multiplexes in each run, and permits typing of a wider range of genetic markers with degraded samples, such as human remains from missing persons' cases. Due to the many benefits of MPS, Battelle and the Ohio Bureau of Criminal Investigation (BCI) have collaborated to validate and implement a customized MPS workflow for use in casework. In this study, Promega's PowerSeq™ Auto/Y prototype amplification kit was selected, along with Illumina's TruSeq® DNA PCR-Free library preparation kits for sequencing on the Illumina MiSeq FGx™ in research-use-only (RUO) mode. Both a developmental and internal validation were performed for this workflow as the Promega PowerSeq Auto/Y kit has not yet been validated by the manufacturer, and MPS laboratory procedures are new to forensic laboratories. Additionally, Battelle's MPS data analysis software, ExactID® with a customized analysis application, proved a helpful tool for handling and evaluating the large amount of informative data produced. Validation guidelines published by SWGDAM were followed as closely as practicable for the MPS study, and the subsequent data from the studies performed confirmed the typing accuracy of the workflow across many variables, including concordance with CE results. The studies performed here show that the PowerSeq System workflow, as validated in the BCI CODIS laboratory, is suitable for integration at a forensic DNA laboratory and valid for processing of casework samples.