

DEVELOPMENT OF A TARGETED NEXT GENERATION SEQUENCING SOLUTION FOR FORENSIC GENOMICS

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Sequencing (NGS) by Synthesis (SBS) enables the entire human genome to be sequenced in one day. Whole genome sequencing (WGS) provides access to all genetic differences between individuals, and is valuable in studying disease and biological systems. While WGS delivers the broadest genomic coverage, it also requires the largest sequencing and interpretation effort. As a simpler alternative, forensic scientists can choose to perform targeted sequencing of PCR products. By sequencing a dense set of forensic loci, casework and database efforts are directed toward the genomic regions that best answer forensic questions, relieving privacy concerns and simplifying analysis. Because it does not depend on allele separation by size, the number of targets interrogated is not limited, allowing a more comprehensive result to be generated.

We describe the development of a targeted amplicon panel for forensic genomics that combines a core of global short tandem repeat markers used routinely today, along with additional forensic loci that can provide information when standard markers would fail to sufficiently resolve a case. Maximizing the number and types of markers that are analyzed for each sample provides more comprehensive and discriminating information for standard samples, as well as challenging samples that contain low quantities of DNA, degraded and/or inhibited DNA, and complex mixtures. The targeted amplicon panel will enable more complex kinship analysis to be performed, and can also reveal phenotypic and biogeographical ancestry information about a perpetrator to assist with criminal investigations. This capability is expected to dramatically improve the ability to investigate dead end cases, where a suspect reference sample or database hit are not available. We will describe the workflow, system, and data analysis tools, and present data from studies with challenging forensic samples, concordance with standard capillary electrophoresis methods, and possible kinship applications.