B133 The Dive Into Next Generation Sequencing (NGS): From Validation to Implementation

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Learning Overview: After attending this presentation, attendees will understand how NGS works and the many different applications for which it can be used. Attendees will also gain insight into the validation and implementation process using this technology.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by providing a demonstration of NGS's ability to augment traditional DNA technology, such as Short Tandem Repeats (STRs). Furthermore, utilizing this technology may assist in providing investigative leads to law enforcement through the use of additional platforms, including genealogy, whole genome mitochondrial DNA (mtDNA) sequencing, phenotypic markers, and ancestry markers.

For almost a quarter of a century, DNA analysis has been dominated by STR and Single Nucleotide Polymorphism (SNP) testing, but it is time to dive into a new era to run in tandem with traditional DNA technology. Utilizing NGS, hundreds of forensic loci are targeted and amplified, including the core locations that have been employed in DNA testing for years. This is important as it allows for compatibility between the current DNA database formats and NGS technology. In addition, NGS can be applied in multiple platforms, which provides the ability to target other sections of DNA, including ancestry markers, phenotypic markers, and whole genome mitochondrial DNA. This technology may heat up cold cases by providing new leads, especially in unidentified remains or unsolved cold cases where a comparable mtDNA profile may establish maternal lineage or genealogy can determine a potential family member leading to identification. With these eye-opening benefits, three different platforms of NGS are being validated and implemented including the Verogen® ForenSeq™ DNA Signature Prep Kit, mtDNA Whole Genome Kit, and a Forensic Genetic Genealogy (FGG) system. The validation studies were designed in accordance with the recommendation of the Scientific Working Group on DNA Analysis Methods (SWGDAM) and the Federal Bureau of Investigation (FBI) Quality Assurance Standards (FBI QAS), including a study that serves to evaluate the effect of processing less samples on a single run in order to obtain more reads per sample. A disadvantage to NGS as opposed to traditional STR testing is the amount of hands-on time that is required by the scientist. To provide a high throughput option for NGS processing, the Aurora Biomed® Versa™ 1000 robotic platform was also evaluated as an alternative to a manual setup. This presentation will review the validation process used to bring online three different NGS platforms as well as the implementation process in the laboratory.

Next Generation Sequencing, Mitochondrial DNA, Genealogy

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