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Arising Issues Implementing Advances in DNA Technology May Tarnish the Gold Standard

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Abstract: Technological advancements in forensic DNA testing are continuously evolving and demonstrate promise to aid in criminal investigations. New DNA processes are being implemented that are more sensitive, software is available to interpret and perform statistics on complex mixtures that historically cannot be resolved, and an individual's phenotype can now be generated along with identity markers. As crime laboratories shift to expanded loci kits, probabilistic genotyping and massive parallel sequencing (MPS), the forensic community must carefully consider the implications and take steps now to responsibly implement these advances.

In 2017, the National DNA Database (CODIS) is expanding from 13 loci to 20 loci. In response, vendors have developed amplification kits that include 24 loci and are also generating full DNA profiles at a sensitivity of 100 pg (~17 cells). Kits now include 10 miniSTRs (loci <200bp) to improve sample recovery and overall achieve an 8 fold increase in the power of discrimination over current methods. Most laboratories do not have experience navigating data generated by miniSTRs or low copy number analysis. While guidelines were issued by SWGDAM in 2014 to address concerns about enhanced detection methods, crime laboratories should acknowledge that the new kits introduce enhanced abilities and be prepared to address this issue.

Mixture interpretation has proven to be difficult, subjective, and variable between humans. Probabilistic genotyping software is allowing the task of interpreting mixtures to be performed by algorithms that can predict outcomes and provide statistical power over current methods. However, if the DNA community does not understand the underlying statistical formulas and cannot come to a consensus on how to interpret mixtures, removing the human element completely creates a black box that may diminish the meaning of a match and the given statistical weight.

Massive Parallel Sequencing is moving the field beyond identity markers to include obtaining information on ancestry, DNA phenotyping, and genetic predispositions. MPS can be powerful and change the way crime is solved but will require careful coordination to properly implement. How the increase in knowledge provided by MPS is used and accepted by legislation and the Court system will steer the course for the future of DNA analysis. Ignoring possible ramification by using this technology for 'investigative leads' only, may be irresponsible considering all foundations used to identify a possible perpetrator may warrant an admissibility hearing.

This presentation will engage the DNA community to discuss the quality issues associated with implementing more sensitive amplification kits, probabilistic genotyping, and MPS. Weaknesses in DNA technological advancements will be identified in order to overcome and improve implementation strategies in the crime laboratory. While the DNA community has the 'power of more' information available now ensuring 'right' information will mitigate potential issues and maintain the gold standard status.