A127 Inter-Tissue Somatic Mosaicism in Blood, Hair, and Epithelial Cheek Cells at the ABI AmpFℓSTR® Identifiler® Loci and Its Effect on Forensic DNA Interpretation

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After attending this presentation, attendees will understand the genetic basis of somatic mosaicism, how it is distinguished from other similar genetic events, how it can be identified and confirmed in a forensic DNA profile, and its significant potential for misinterpretation when comparing DNA profiles of more than one biological source from one donor.

The presentation will impact the forensic science community by helping forensic scientists predict how often they are likely to encounter somatic mosaics in routine casework and whether their frequency is high enough to warrant changes in their procedures for comparing reference samples from suspects with crime samples and collecting samples for NDIS/CODIS database. Even if the frequency is low, several somatic mosaics are likely to be present in NDIS (>10.5 million offender profiles) or CODIS (>2 million offender profiles). The ever-growing CODIS database is currently based on inputting DNA derived from only on tissue origin. The detection of inter-tissue somatic mosaics in this study would suggest that forensic scientists should consider collecting blood, hair, and saliva samples from each offender. This becomes even more important if a partial profile from a crime scene is used to search the database. In addition, past exclusions (particularly those based on one or only a few mismatches) will need to be reassessed, possibly leading to new leads, arrests, and convictions for crimes that are currently unsolved.

Crime solving using DNA relies on the assumption that each bodily tissue of an individual exhibits the same DNA profile. Based on this prevailing assumption, positive identification of a suspect or victim can be made by matching the crime scene sample to a reference sample regardless of the origin of the bodily tissues used for the comparison. For example, a hair found on a ski mask left at a crime scene is assumed to have the same DNA profile as a blood reference sample collected from the person who left the hair. Although this assumption of inter-tissue identity is generally sound, it is not always correct because genetic mutations can cause differences in inter-tissue profiles. Somatic mosaicism is caused by mutations during embryogenesis that lead to differences in the genetic make-up of different tissues in the adult. Differences can manifest in several different ways, depending on how early the mutation occurred and whether the mutation created an allele that the individual did not already carry.

Inter-tissue somatic mosaics can be detected by collecting tissue samples from different parts of a person's body and comparing their Identifiler® profiles. The Identifiler® loci are Short Tandem Repeats (STRs), which tend to mutate more rapidly than other regions of the human genome. Therefore, it is more likely for a person to exhibit mosaicism at these loci than at most other loci in the genome. Somatic mosaicism has been well-researched in the medical community, but few studies have been completed for the benefit of forensic DNA identification.

This is the first forensic-related, multi-year research on somatic mosaicism that incorporates three distinct tissues for analysis (blood, hair, and epithelial cheek cells) from multiple donors. Currently, one out of 352 donors have been identified as a somatic mosaic. The donor's blood and saliva displayed a tri-allelic pattern (11, 12, 13) at the D8S1179 locus. The donor's hair sample also showed a small peak at the 12 allele; however, the allele was not included, because it did not cross the peak height threshold set at 20%. The results of this study indicate that somatic mosaicism can be easily detected as a tri-allelic pattern. Based on the findings, the rate of somatic mosaicism is very low (one out of 352 donors), but several somatic mosaics are likely to be present in NDIS/CODIS.

It is assumed that the DNA profile from a blood drop found at a crime scene can be successfully used to search the CODIS database, even though most of the database samples are generated from a single source (buccal cells). The detection of inter-tissue somatic mosaics in this study would suggest that forensic scientists should consider cross-tissue comparison from each offender. Further research with larger population sizes would benefit the interpretation of DNA casework.

Somatic Mosaicism, Genetic Mutation, DNA

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