INTER-TISSUE SOMATIC MOSAICISM IN BLOOD, HAIR, AND EPITHELIAL CHEEK CELLS AT THE ABI AMPFLSTR® IDENTIFILER® LOCI AND ITS EFFECT ON FORENSIC DNA INTERPRETATION

Katerina G. Doneva, B.S., Shannon Sheehan, M.S., Lauren Gaggero, M.S., Ron Moat, M.S., and Ruth E. Ballard, PhD.
University of California Davis, Forensic Science Masters Program, 1333 Research Park Drive, Davis, CA 95616, USA

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. Promega Corporation has posted an article on their website of a study in which 4 separate occurrences of somatic mosaicism in 190 hair samples were detected (Silvia et al). 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, 1 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 and doesn't have an effect on the "major" genotype of an individual. The rate of somatic mosaicism is very low (1 out of 352 donors), but several somatic mosaics are likely to be present in 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. ⌘