A25 Interesting Case - XO Male in Forensic Casework

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After attending this presentation, attendees will become familiar with an unusual STR genotype for a male individual and the ways in which male quantification and Y-STR typing can be used to aid in the confirmation of male DNA.

This presentation will impact the forensic community by providing education and a case study on a forensic casework sample that appeared genotypically female but was actually male. Attendees will observe how male quantification and amplification using Y-STRs can help resolve apparent discrepancies such as these.

A homicide case was submitted to the Harris County Institute of Forensic Sciences-Forensic Biology section for analysis. The case involved two male defendants charged with capital murder for the beating and shooting death of a victim during a convenience store robbery. Swabbings taken from a bat used during the commission of the crime and swabs from the counter area where the crime occurred were

presumptively positive for blood. DNA analysis was performed on these samples and the DNA profiles obtained were compared to the known reference samples from the defendants and the victim. Mixtures of DNA were obtained from the bat and two of the counter area samples which were consistent with the reference from the victim (major contributor). The suspects were excluded from these mixtures. Single source DNA profiles were obtained from two of the counter area samples that were also consistent with the reference sample from the victim.

The profiles from the crime scene, which were consistent with the victim, yielded an “X” at the Amelogenin locus. Since the decedent in this case was male, a genotype of “XY” was expected at the Amelogenin locus. Three explanations for this discrepancy were explored: (1) the absence of a “Y” chromosome was due to the fact the victim was female;

(2) deletion on the Y-chromosome; and, (3) mutation in the primer binding site of the AMEL locus on the Y-chromosome of this individual. The autopsy of the victim also had been performed; therefore the medical examiner on the case was contacted for more information. The medical examiner confirmed that the victim was physically male.

The Forensic Biology section had validated and implemented combined human/male DNA quantification and Y-STR analysis during the tenure of this case. It was decided to use these DNA analysis tools to resolve the issue of the discrepancy of the sex of the victim. The commercial human/male quantification kit used determines the amount of human DNA in a sample and also utilizes a male probe that targets the sex determining region of the Y chromosome (SRY) to determine the amount of male DNA present in a sample. Male DNA was detected at quantification. To determine whether a region of the Y-chromosome had been deleted or a point mutation in the primer binding site of AMEL was the culprit, Y-STR testing was performed on the victim’s standard. A full Y-STR profile was obtained from the victim’s reference sample with the exception of DYS458 which is the closest locus to the AMEL locus of the Y-STR loci tested.

Literature research documented that Amel Y and DYS458 are adjacent to each other on the Y chromosome (6.79 Mb and 7.92 Mb, respectively). Therefore, a deletion occurring within this area could affect the detection of both loci. Publications have also documented a 3.2-3.6% sex test failure rate in individuals of Indian descent; the victim in this case was of Indian descent. This case study demonstrated the advantages of a quantitation system that determines both human and male DNA and the utility of Y-STR analysis to provide a comprehensive analysis of genetic information in casework samples.

Amelogenin, Y-STR, STR

 ** Presenting Author*