B159 A More Sensitive Sex Determination Assay

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After attending this presentation, attendees will learn of the utility of human DNA primers that target multicopy nuclear sites on the Y chromosome and autosomes for determining sex in samples of forensic relevance, particu- larly those for which standard genetic sexing techniques fail.

This presentation will impact the forensic community and/or humanity by addressing a means of determining the sex of forensic samples that contain degraded DNA and/or DNA that is present at extremely low quantities, which typically provide a challenge to crime laboratories. This presentation will demonstrate the utility of multicopy nuclear targets to determine the sex of forensic samples, thus assisting the criminal investi- gation process.

DNA analysis is a very useful tool in the criminal investigation process; however, analysis can be complicated by a lack of viable DNA. Aged skeletal material, blood, and tissues, or shed hairs, nails, and skin cells, often do not yield DNA amplification products when single copy nuclear loci, those typically tested by the crime laboratory, are examined. Sex determination of a forensic sample is generally made using the single copy gene amelogenin. If this marker cannot be amplified owing to low DNA quantity, it is still possible that amplification of high copy number loci may be successful. In theory, if loci are present as many copies on the chromosome and small amplicons are targeted, amplified products can be obtained with extremely low starting concentrations of DNA. These products can then be analyzed using standard agarose gel or capillary elec- trophoresis.

In the experiments described here primers for the Y chromosome locus DYZ1, which exists at 2000 to 4000 copies per cell, were designed and used to test control DNA at decreasing concentrations, as well as foren- sically useful samples (*e.g.*, bone and hair shafts). As a positive control, primers for the chromosome 17 alpha satellite DNA, which exists at approximately 1000 to 2000 copies per cell, were designed and tested. Results show that the multi-copy markers are two or more orders of mag- nitude sensitive than standard amelogenin amplification when control DNA is tested. Likewise, in many cases forensically relevant samples can be sexed using this method when amelogenin testing fails, while in con- trast, amelogenin analysis was not successful when the high copy markers were negative. Overall, the ability to yield information using the high copy number markers on samples that could not otherwise be amplified will be a useful tool for the criminal investigative process.

This project was supported in part by Grant Number 2002-IJ-CX- K016 awarded by the National Institute of Justice, Office of Justice Programs, US Department of Justice. Points of view in this document are those of the authors and do not necessarily represent the official position or policies of the US Department of Justice.

Sex Determination, DNA, Multicopy Loci

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