B37 The Evaluation of the Effects of Linked Markers on Kinship Testing

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After attending this presentation, attendees will better understand the implications of linked loci in kinship calculations and how best to approach such loci in familial relationship testing.

This presentation will impact the forensic science community by demonstrating to what degree three sets of linked loci (vWA and D12S391, CSF1PO and D5S818, and SE33 and D6S1043) are linked and by illuminating the potential impact of treating these as independent instead of linked. The results of this research may impact the processes involved in familial kinship testing for both domestic and foreign immigration cases by more accurately depicting pedigrees.

Kinship testing cases are common practice and important in many immigration cases globally. The kits used to analyze the DNA in such cases vary, but very often will contain linked markers. Linked markers are used occasionally in forensic DNA analyses, including kinship calculations, often with little regard for their potential impact on the calculations. The simple product rule should not be used with linked markers that are shown to not segregate independently. What should be considered is the diplotype frequency with its own frequency for recombination based on a diplotype database.

DNA from previously extracted samples from 96 families with multiple children (both full and half sibling children) from four ancestry populations (European, Asian, Hispanic, and African) from past paternity cases were supplied by The George Washington University, Department of Forensic Science, sourced from the Applied Genetics Technology Corporation (AGTC) in Denver, CO. These samples were quantified using Quantifiler® Duo, amplified using GlobalFiler™ and VeriFiler™ Direct PCR amplification kits, run on an Applied Biosystems® 3130 Genetic Analyzer, and typed using GeneMapper IDX® software version 1.4.

The results of the genetic typing were exported into Excel® where allele and diplotype frequencies were calculated for each population of n=24 families per population. Additionally, recombination frequencies were calculated at each linked diplotype that were further utilized to determine if they exhibited linkage disequilibrium.

To demonstrate the impact of using these loci in conjunction with the product rule for kinship cases, familial relationships likelihoods were calculated assuming independent assortment of the genes. These relationship likelihoods were then compared to relationships calculated by accounting for linkage to show the true impact of not accounting for linked loci.

This presentation will discuss the level of linkage between vWA and D12S391, CSF1PO and D5S818, and SE33 and D6S1043, demonstrate the impact of ignoring the linkage disequilibrium between the loci pairs, and discuss more efficient ways of approaching the use of linked loci in familial testing.

Research Funded by The Department of Homeland Security.

Kinship, Linked Markers, Diplotypes

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