A159 Determining the Evidential Strength of mtDNA and Y-STR Matches

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After attending this presentation, attendees will be aware of methods to determine the numerical strength of lineage marker profiles.

This presentation will impact the forensic science community by introducing an improvement on existing methods of giving the numerical strength of lineage marker profiles.

The interpretation of mtDNA and Y-STR evidence differs from autosomal DNA largely because these two types of DNA are inherited uniparentally and without recombination. The usual method for interpreting such markers, refereed to collectively as lineage markers, has centered around the empirical count in a database. This is termed the counting method. Areas of current debate relate to the assessment of sampling uncertainty in such a count and methods to deal with subpopulation effects.

Sampling uncertainty is often assessed using the method of Holland and Parsons.[1] This method assumes normality and Holland and Parsons recognized that such an assumption would not be appropriate for low frequencies. However, the method has not been refined in the 10 years since publication. In this paper we present a standard frequentist approach, known since 1934,[2] and a Bayesian approach that remove the difficulties associated with non-normality. Trials with these two methods confirm that the Holland and Parsons method is inaccurate, as suggested by the initial authors, not conservative, and should be replaced.

Lineage markers are known to show strong subpopulation effects.[3] As such it is expected that a general population database count may not be applicable to a more localized subpopulation. However, the application of the known subpopulation correction appears extreme. The known formulation would change the database count *f* to . Here is the coancestry coefficient that is often assessed as being of the order of 0.02 - 0.10 for lineage markers so that such a correction would completely dominate the frequency term. However, although variation between subpopulations is large, variation within subpopulations is also large if the full haplotype is utilized suggesting that from single loci may overstate the differentiation. Recently Budowle et al[4] recognized this and estimated for the full haplotype utilizing the method of Weir and Cockerham[5] which will not produce reliable estimates for such sparse data. Another approach, that of Ewens[6], does appear applicable and this suggests that, indeed, estimates from single loci are misleading and that much lower estimates of may be sustainable for multilocus lineage marker haplotypes, as envisaged by Budowle et al.

References:

1 Holland, M.M. and T.J. Parsons, *Mitochondrial DNA Sequence Analysis – Validation and Use for Forensic Casework.* Forensic Science Review, 1999. 11(1): p. 21-50.

2 Clopper, C.J. and E.S. Pearson, *The use of confidence or fiducial intervals illustrated in the case of the binomial.* Biometrika, 1934. 26: p. 404-413.

3 Irwin, J.A., et al., *Development and expansion of high-quality control region databases to improve forensic mtDNA evidence interpretation.* Forensic Science International: Genetics, 2007. 1(2): p. 154-157.

4 Budowle, B., et al., *The effects of Asian population substructure on Y STR forensic analyses.* International Journal of Legal medicine, 2009. 11: p. 64-69.

5 Weir, B.S. and C.C. Cockerham, *Estimating F-statistics for the analysis of population structure.* Evolution, 1984. 38: p. 1358-1370.

6 Ewens, W., *The sampling theory of selectively neutral alleles.* Theoretical Population Biology, 1972. 3: p. 87-112.

mtDNA, Y-STR, Lineage Markers

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