A155 How Y Haplotypes Are Distributed and How They're Not

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After attending this presentation, attendees will have a mental picture of the patterns of groups of men who share Y haplotypes. This understanding is important for giving a realistic appraisal of the evidential strength of a Y haplotype forensic match.

This presentation will impact the forensic science community by simplifying and clarifying how to understand the evidential value of haplotype evidence — mtDNA as well as Y. It will not solve the problem of coping with population inhomogeneity, but will provide promising illumination for the way forward.

Forensic evaluation of DNA evidence rests in part on understanding the underlying population genetics — how the DNA is distributed in the population. This is especially true for lineage markers including Y haplotypes. It is natural, but wrong, to suppose that the familiar autosomal tools and principles adapt to the Y domain; they don't. Obviously, the autosomal product rule is lost, and there are important but less obvious examples as well. In particular, while sample frequency is a fair approximation to autosomal allele matching probability, for today's Y haplotypes of many STR loci, it's absurd. The common expression "unrelated man" is sometimes a good enough approximation for traditional work; however, since a patrilineal connection without intervening mutation is almost the only possibility for two men to share a Y haplotype, reasoning by analogy with autosomal DNA is to dramatically misperceive the evidence. The random mating/product rule approach to autosomal calculation using allele probabilities can be refined by taking into account the "inbreeding coefficient" θ, the chance two alleles have a common ancestor without mutation. The Y haplotypes situation is the opposite where one starts with θ and it is 97% of the story (for YFiler®) and anything else is a minor refinement.

Alleles at neighboring autosomal STR loci replenish one another by step-wise mutation as is obvious from observing the compact and sometimes unimodal distribution of allele frequencies. It's natural to suppose that haplotypes behave similarly, but they don't. Mutation along a patrilineage away from a type, then mutation back to the original type is an insignificant phenomenon. To a close approximation, all the men in the population with any particular haplotype are men descended from a common ancestor (typically 500 years past) without mutation. The size of such a group is typically thousands or tens of thousands of men, all mutual cousins. Understanding how those cousins are distributed in the world is how to understand haplotype evidence.

Y-Haplotype, Lineage Marker, Population Genetics

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