B156 Median Network Analysis of mtDNA Haplotypes in the Basques of Southern Idaho

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After attending this presentation, attendees will gain an appreciation of the role of genetic drift (and specifically the founder effect) as it affects genetic variation and haplotype frequency in a specific sub-population, the immigrant Basque population of southern Idaho. Attendees will learn how median network representations of mutational differences between haplotypes can be used to infer the genealogical relationships among sequences.

This presentation will impact the forensic community and/or humanity by demonstrating that through migration and settlement of human populations, the regional pattern of genetic profiles can be explained as an admixture of specific lineages of the founding populations. Genetic profiles of the population are estimated in this study by mitochondrial DNA sequences of the first and second hypervariable segments (HVS1 and HVS2).

This non-coding region of mtDNA is known to have a much higher mutation rate than nuclear DNA, and thus lineage-specific mutations can be used to characterize sub-populations, and in some cases even identify individuals (for example, the remains of the Romanov family). The specific objectives of this study are as follows:

1) create a median network representation of Basque HVS sequences and their mutational distances from each other (haplotype network)
2) infer genealogical relationships between Idaho and European Basque maternal lineages

The Basque are a culturally and linguistically distinct sub- population within Europe. Characterized by a long-standing independent political history, and a unique language (not known to be related to any other extant language), the Basque people have long been of interest to scholars studying the genetic diversity of Europeans.

To collect samples for this study, individuals of Basque heritage from Idaho volunteered a buccal swab, from which mtDNA was extracted using the Qiagen Mini-Amp DNA kit. The hypervariable segments of the mtDNA control region were then PCR amplified, and sequenced using a LICOR 4300 DNA Analyzer. To ensure samples were from people of maternal Basque heritage, a questionnaire protocol was followed: each volunteer filled out a questionnaire, and the maiden names and birthplaces of their mother and their mother's mother were recorded. Only samples that have a recognizably Basque maiden name for their maternal grandmother were considered for this study. A median network was created using the program Network 4.0, distributed by Fluxus Engineering.

mtDNA, Haplotype Frequencies, Median Network