A188 Determination of Ancestry in Forensic Skeletal Cases From Haplogroup Assignments Based on Hypervariable Region Mitochondrial DNA Sequence Data

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After attending this presentation, attendees will understand the circumstances in which mtDNA can potentially help to determine the ancestry of skeletal remains and the limitations of any such determination.

This presentation will impact the forensic science community by informing forensic anthropologists and geneticists of how their disciplines can jointly inform the determination of ancestry of unknown skeletal remains.

In forensic casework, DNA is primarily used for the individuation of evidence or the identification of remains. This is especially true for autosomal DNA (auDNA), which can be used in a positive identification, but is generally the case for Y-chromosome (yDNA) or mitochondrial DNA (mtDNA) as well. While variation in yDNA and mtDNA actually only places an individual within one or more particular groups of paternally or maternally related individuals, it is generally used in conjunction with other circumstantial evidence (such as a passenger manifest) to determine individual identity. When individuation is not possible, either because references are not available for comparison or because no victim names are known, how well can DNA be used to assign an individual to a population group?

MtDNA is the type most frequently sequenced from skeletal remains, because of its high copy number and good preservation. Because of its haploid, or unilateral, inheritance, mutations in mtDNA over time have led to a branching tree of variation, where any specific haplotype, or genome, can be placed in relationship to all others. Branches of this tree are referred to as haplogroups, each of which groups various haplotypes united by common descent. While early studies of mtDNA variation at the population level focused on coding region mutations, using these to define the original haplogroups, forensic applications generally sequence the hypervariable region (HVR) and occasionally the broader control region. The high mutation rate across the HVR makes it iDrug Enforcement Administrationl for individuation, but also means that a particular mutation may well have occurred multiple times and not necessarily signify anything about relatedness. Individuals belonging to different haplogroups, and exhibiting different polymorphisms elsewhere in their mitochondrial genome, may bear the same sequence in the HVR. Some polymorphisms in the HVR are more stable and are associated with particular haplogroups, but most haplogroups are defined on the basis of markers that are not sequenced in standard forensic casework.

To use mtDNA for determining ancestry in a forensic setting, three questions must be addressed: (1) how accurately can a given sequence be assigned to a specific haplogroup; (2) how accurately can a given haplogroup be assigned to a specific ancestral population; and, (3) what is the correlation between that ancestral population and a modern “racial” category?

Comparison of HVR sequence data to the global mtDNA phylogeny allows many sequences to be accurately placed within broad haplogroups, and some to be much more specifically sorted. Others, particularly within the M and R macrohaplogroups, cannot be accurately assigned on the basis of HVR data alone. Even in such cases, however, the range of possibilities may be broadly restricted to a given continental origin.

Because of its haploid inheritance, mtDNA only reflects an individual’s maternal ancestry through previous generations. Population genetic studies have found that it has a very weak correlation with geography on a local level, because of the movement of wives between communities. On a broader, continental level, it does have a high correlation with geography. That is, most haplogroups, or groups of sequences or haplotypes united by shared descent, tend to be found in populations originally from one continent or another.

Historically, most gene flow occurred at the sub-continental level. Over the past few centuries, individuals and entire populations have moved over much broader distances, creating a higher degree of gene flow. In a forensic context, this means that any ancestral determination based upon genetics has to take the context of the remains into consideration and has to explicitly address the question of mixed descent and cultural classification.

As an example, the very diverse macrohaplogroup L, aside from its descendant clades M and N, is restricted in distribution to African populations. In forensic casework, an individual of haplogroup L can be reliably stated to be of African maternal ancestry. But does this always correlate with Black, or African-American? In a sample of U.S. military casework, most occurrences of haplogroup L were indeed individuals who were racially classified as Black; however, several were White. It is most likely that these were not “mixed race” individuals, defined as those with parents or grandparents of distinct ancestry; rather, they were individuals descended, as far as they knew, from uniformly White ancestors, but who had a distant maternal ancestor who was African-American. These same individuals were classified as White by physical anthropologists, in accordance with their own self-categorization.

Another example is provided by World War II-era skeletal remains recovered from the Pacific theater.

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Because there was minimal historic gene flow between Japan and Europe, while the limited migration from Japan to the United States had occurred within one or two generations before the war, in most cases the haplogroup determined from a given mtDNA sequence allows a set of remains to be assigned to either Japanese or American origin.

Because race is a cultural category, albeit one based upon perceived biological differentiation, it will never perfectly correspond with determinations of ancestry based upon biological variation. Nonetheless, mtDNA variation can help in hypothesizing the ancestry of a given set of remains and what race their owner might have been classified in, if its limitations are clear.

The views expressed herein are those of the author and not necessarily those of the Joint POW/MIA Accounting Command or the U.S. Department of Defense.

Mitochondrial DNA, Ancestry, Human Skeletal Remains

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