A132 A Review of the Changing Use of Mitochondrial DNA in Identifications by the Central Identification Laboratory

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After attending this presentation, attendees will understand how the Central Identification Laboratory and Armed Forces DNA Identification Laboratory use mtDNA to identify United States service members lost in historic conflicts, and how mtDNA can be used in different ways as the evidence and reference database sizes increase.

This presentation will impact the forensic science community by showing the power of large mtDNA evidence and reference databases for directly making identifications and for constructing hypotheses that may lead to identifications.

In 1991, the U.S. Army Central Identification Laboratory—Hawaii (CILHI) made its first identification that used mitochondrial DNA (mtDNA), followed by three more in 1994 (Holland et al. 1993).[1] After questions were raised about the reliability of mtDNA-based identifications, the Defense Science Board Task Force on the Use of DNA Technology for the Identification of Ancient Remains conducted its own review. Their 1995 final report recommended that mtDNA could indeed be used for the identification of the remains of U.S. service members lost in prior conflicts. Over the following four years, mtDNA reports issued by the Armed Forces DNA Identification Laboratory (AFDIL) supported 36% of the identifications made by the CILHI. In 2000, for the first time, mtDNA reports supported a majority of identifications, and since then 79% of identifications by the CILHI and its successor organization the Joint POW/MIA Accounting Command—Central Identification Laboratory (CIL) have been supported by mtDNA.

The manner in which the CIL uses mtDNA results has developed and expanded over time. The earliest identifications resulted from comparisons of a single evidence sequence with a maternal family reference sample (FRS) obtained for a single casualty by that casualty's respective military service. These were followed by comparisons between one or more evidence sequences and a small, closed population of casualties. In the late 1990s, as CILHI began analyzing remains recovered from North Korean battlefields, potential casualty populations became more open. As a result, the services were asked to obtain maternal references for much longer lists of casualties. In 2003, the CIL began submitting megacomparison requests to AFDIL, asking for a long list of potential casualties to be compared to a list of cases recovered from a given region. At the same time, it became clear from mtDNA results that the remains of single individuals might be spread across multiple accessions from North Korea. To address the question of commingling, AFDIL began generating spreadsheets that charted the occurrence of each individual sequence across all of the different cases received from North Korea. In 2008, this spreadsheet was extended to include all those remains recovered by the CIL in North Korea. Most recently, the same tabulation has been done for all remains associated with the Vietnam War. At the same time, the proportion of casualties for whom FRS are available has increased to 68% for Korea and 69% for Vietnam. Given the completeness of this reference set, it now makes sense to run each distinct evidence sequence against all the FRS available for that conflict.

At the same time that the FRS database has grown, so has the reference population database. The first mtDNA comparisons by AFDIL relied upon a reference population of 715 individuals reported in the literature. In 1998, AFDIL began using what is currently the SWGDAM database, which by 2002 contained 4839 individuals. In 2007, AFDIL began using its own Casework Population Database (CPD) of 10,428 individuals, including samples from each racial and continental grouping. Even when an evidence sequence does not match any FRS, if it is present within the CPD, hypotheses can be generated about the ancestry of the individual. This has allowed the CIL to focus attention on these remains exhibiting sequences consistent with individuals of probable American ancestry (whether European, African, or Native American), as opposed to those indigenous to Korea, Southeast Asia, or elsewhere in Asia or the Pacific. In cases from Europe, such determinations are clearly of less value; however, in at least one case, greater credence could be given to a claim that a unilaterally turned over bone derived from an American aircraft loss because the mtDNA sequence obtained was only found in the CPD among U.S. Hispanics.

MtDNA is, in theory, an exclusionary method of identification: If an evidence sequence does not match an FRS, then the sequence does not represent that casualty. If a sequence does match an FRS, then it is possible, but not certain, that the sequence represents that casualty. In small, closed populations, all individuals generally have different sequences, so this is not a problem. In large scale comparisons, such as that between hundreds of evidence sequences and thousands of FRS, many sequences may represent multiple skeletal individuals and match multiple casualties. In these comparisons, mtDNA is a source of hypotheses, which can then be tested using other methodologies. If, for instance, a sequence from Korea or Southeast Asia matches only one casualty from that conflict, then the identitity of that sequence with that casualty is a fairly strong hypothesis. If circumstances of recovery are consistent with those of loss, and dental and anthropological comparisons are also consistent, identification may be possible. The first such IDs were made in 2004 and 2005, when skeletons turned over by the Vietnamese and North Koreans, respectively, were identified based on unique blind mtDNA matches combined with biological profiles of the skeletal remains and odontological analysis of the dental remains. If a sequence matches multiple casualties, or has been obtained from multiple sets of remains, anthropology and odontology may be insufficient to support an identification. AFDIL is now developing the capability to use Y-DNA and autosomal STR comparisons to support mtDNA matches, and both methods have been used in recent identifications (Irwin et al. 2007).[2] The views expressed herein are those of the authors and not necessarily those of the Joint POW/MIA Accounting Command, the Armed Forces Institute of Pathology, the U.S. Army Surgeon General, nor the U.S. Department of Defense.

References:

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2. Irwin J, Edson S, Loreille O, Just R, Barritt S, Lee D, et al. The intersection of genetic identity: the application of multiple marker systems and new technologies to establish identity 50 years after death. J Forensic Sci 2007;52:1115-8.

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