A105 Estimating Ancestry and Region of Origin: A Multidisciplinary Approach to the Identification of Unknown Individuals

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Learning Overview: After attending this presentation, attendees will better understand the benefits and limitations of using free, publicly available mitochondrial DNA (mtDNA) databases to complement visual and anthropological ancestry estimates in cases of unidentified individuals.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by demonstrating how mtDNA ancestry predictions perform in comparison to visual and anthropological ancestry estimates. In addition, the mtDNA databases will be assessed for their utility in making predictions on region of origin, which could be especially useful in the identification of migrants.

Forensic anthropologists often collaborate with other specialties to assist in the identification of unknown individuals by contributing biological profiles, as well as by providing investigative resources, maintaining oversight of long-term unidentified individuals, and by suggesting innovative scientific approaches. The purpose of this study is: (1) to determine the accuracy of ancestry estimates from mtDNA databases and assess their utility in building upon ancestry estimates (both anthropological and visual), and (2) to determine their efficacy in estimating an individual's region of origin, especially in individuals who are potentially migrants. This research also highlights the benefits of collaboration between the forensic science disciplines, as demonstrated by the Forensic Anthropology and Forensic Biology Units at the New York City Office of Chief Medical Examiner (OCME).

The test sample consists of 41 OCME cases of identified (20) and unidentified (21) individuals for whom an mtDNA profile was developed. The sample consists of 19 cases in which ancestry was visually assessed and 22 cases in which ancestry was anthropologically estimated using macromorphoscopic traits and craniometrics. An additional 11 OCME staff members were included for quality control measures.

The mitochondrial sequences of each individual were searched through the European DNA Profiling Group (EDNAP) mtDNA Population Database (EMPOP) mtDNA online database, v3/R11. The haplotype lineages and region of origin predictions produced by EMPOP were then compared to the ancestry estimates. All mtDNA sequences and anthropological ancestry estimates were generated by the Forensic Biology and Forensic Anthropology Units at the OCME.

Results revealed 83% (34/41) overall agreement between the haplotypes derived from the EMPOP database and the visual and anthropological estimates of OCME cases. Comparison of the mtDNA results with visually assessed ancestry revealed 89% agreement (17/19), while comparison with anthropologically derived ancestry estimates revealed 77% agreement (17/22). Of the anthropology cases in which the two methods yielded conflicting ancestral predictions, approximately 40% exhibited incomplete/damaged crania and 40% exhibited morphoscopic traits/craniometrics consistent with multiple ancestral groups. For all known/identified individuals, 6 of 7 anthropological ancestry estimates and 19 of 20 haplotype predictions overlapped with the individual's confirmed ancestry. Individuals of confirmed "Hispanic" and "Middle Eastern" ancestry may exhibit either African, Asian, or European haplotypes and, therefore, any of these haplotypes would be considered overlapping with a "Hispanic" or "Middle Eastern" visual/anthropological assessment. An additional 11 OCME staff members were tested for quality control measures, and 9 of these individuals exhibited haplotypes that matched their confirmed ancestry.

The OCME dataset includes 8 individuals with known region of origin, which was confirmed through discussions with families and foreign consulates during the identification process. The haplotype density maps produced by EMPOP overlapped with the region of origin in 6 of the 8 cases. A quality control test of 11 OCME staff members demonstrated overlap in regions highlighted on the map in only 6 individuals. Maps for individuals with European and African maternal lineages were diffuse and non-specific, while those of Asian maternal lineages, AfroCaribbean, as well as "Hispanic" lineages from Central and South America were predicted with greater specificity.

Results indicate that mtDNA haplotype predictions could be a useful complement to the anthropological or visual ancestry assessment. These databases are free and publicly available and, with the collaboration of forensic biology and anthropology experts, could provide additional investigative leads. However, since mtDNA only reflects the maternal lineage and ancestral self-identification is complex, it is not recommended to use these databases without collaboration with anthropologists and biologists. In certain cases, the EMPOP database density maps can add additional information that is informative to the identification of potential migrants, especially those of Asian or Central and South American origins. Preliminary results indicate that Y-chromosomal Short Tandem Repeat (Y-STR) databases also show promise in predicting ancestry and region of origin for unidentified male individuals.

Ancestry Estimation, Mitochondrial DNA Ancestry Database, Unidentified Remains