

PHYLOGENIC CHARACTERIZATION OF THE SWGDAM FORENSIC mtDNA DATA SET

Mark R. Wilson M.A.¹, Marc W. Allard Ph.D.², Kevin Miller Ph.D.³, Keith Monson Ph.D.¹, Bruce Budowle Ph.D.³

¹Federal Bureau of Investigation, Counterterrorism and Forensic Science Research, Quantico, VA

²George Washington University Department of Biological Sciences, Washington, DC

³ Federal Bureau of Investigation, DNA Analysis Unit II, Washington, DC

Currently, the Scientific Working Group on DNA Analysis Methods (SWGDAM) mtDNA data set is used to infer the relative rarity of mtDNA profiles (i.e. haplotypes) obtained from evidence samples and for identification of missing persons. The Caucasian haplogroup patterns in this forensic data set have been characterized using phylogenetic methods. The assessment reveals that the data set is relevant and representative of U.S. and European Caucasians. The comparisons carried out were both the observation of variable sets within the control region (CR) and the selection of a subset of these sites which partition the variation within human mtDNA control region sequences into cluster (i.e. haplogroups). The aligned sequence matrix was analyzed to determine both single nucleotide polymorphisms (SNPs) in a phylogenetic context, as well as to check and standardize haplogroup designations with a focus on determining the characters that define these groups. To evaluate the data set for forensic utility, the haplogroup identification and frequencies were compared with those reported from other published studies. Similar studies on African-American, Hispanics, Asian, and Native American populations are currently underway. The preliminary results show consistency with other published datasets, further validating the forensic population database.