



A96 The Detection of Genetic Information in the Enamel Proteome

*Caleb Kiesow**, United States Air Force Academy, Colorado Springs, CO; *Katelyn Mason, PhD*, Lawrence Livermore National Laboratory, 7000 East Avenue, Livermore, CA 94550; *Deon Anex, PhD*, Lawrence Livermore National Laboratory, 7000 East Avenue, Livermore, CA 94550; *Bonnee Rubinfeld, MSc*, Lawrence Livermore National Laboratory, 7000 East Avenue, Livermore, CA 94550; *Laura A. Regan, PhD*, Office of Net Assessment, 1920 Defense Pentagon, Rm 3A932, Washington, DC 20301-1920; *Bradley Hart, PhD*, Lawrence Livermore National Laboratory, 7000 East Avenue, Livermore, CA 94550; and *Glendon Parker, PhD**, Protein-Based Identification Technology, 4421 Ashwood CMN, Fremont, CA 94538

After attending this presentation, attendees will better understand alternative scientific and statistical approaches for calculating measures of human identity from teeth.

This presentation will impact the forensic science community by informing attendees that genetically variant peptides from tooth enamel can potentially be used to infer the status of Single Nucleotide Polymorphism (SNP) loci in a subject's genome. The past few decades have seen the impact of nuclear and mitochondrial DNA typing on forensic science. The method discussed in this presentation introduces a third identifying method based on protein typing.

Human identification frequently depends on the ability to extract genetic information from human remains. In most cases, DNA typing is sufficient to statistically link an individual with biological material. In the event that DNA is absent or degraded, additional identification methods are required. Given the increased need for laboratory-based quantitative forensic methodologies, genetic variation in protein is an attractive alternative to develop novel identification technologies.

Enamel is the most robust human tissue and persists for long periods in the environment. The calcium apatite crystals encase and protect endogenous protein that contains genetic information in the form of single amino acid polymorphisms, the result of non-synonymous SNPs. This type of genetic variation is distributed across many populations and can be used to develop measures of human identification.

Blocks of enamel (20mg) were obtained from four subjects and were milled and dissolved in HCl for 1h at 56°C in the presence of reductant. The resulting supernatant was pH neutralized, carboxymethylated, and digested with trypsin protease in the presence of mass spectrometry-compatible surfactant. The resulting peptide mixtures were applied to a Thermo™ Q Exactive™ plus hybrid Orbitrap/linear ion trap mass spectrometry instrument. The resulting proteomic datasets were analyzed by using the "Global Proteome Machine" peptide spectra matching algorithm (www.thegpm.org). The peptide datasets were screened for previously characterized and genetically validated variant peptides. Six genetically variant peptides were observed in the enamel proteome across the four subjects and originated from five proteins, including COL1A2, one of the two major forms of collagen in teeth and bones.

This data indicates that genetically variant peptides in enamel are a potential and orthogonal means to develop statistical measures of human identification in the event of compromised, degraded, or absent DNA. This will have particular relevance for skeletal remains in which DNA is either degraded or absent. In the context of mixed skeletal assemblages, profiles of genetically variant peptides may also provide information about biodistance between different skeletal components.



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