RECOVERY OF MITOCHONDRIAL GENOME FROM CREMATED HUMAN REMAINS

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In cases of fires or explosions, forensic identification by traditional methods such as fingerprinting or anthropological analysis is often not possible. DNA identification of charred or incinerated remains may be the last resort for family members wishing to identify their loved ones. The recovery of DNA from burned human remains has been notoriously difficult. Some studies have been successful in typing DNA from charred remains and dental pulp, however, concerns exist regarding the quality and purity of DNA that is recovered. Nuclear DNA in bone is often limited and PCR inhibitors can impede amplification of short tandem repeats (STRs). The circular structure and subcellular sequestration of mitochondrial DNA (mtDNA) may provide some protection from degradation, and high copy number of mtDNA per cell increases sensitivity of DNA typing assays making it better-suited for DNA identification of severely degraded samples such as burned remains.

In this study, we sequenced mtDNA from an individual whose remains had been incinerated in a commercial crematorium. We extracted DNA from both ash and small bone fragments and quantified mitochondrial genome copies via qPCR assay. Hypervariable region 1 (HV1) was amplified and sequenced via Sanger sequencing. Whole mtGenome was amplified using a multiplex PCR assay, libraries prepared and sequenced via MPS.

We compared HV1 and whole mtGenome sequences to confirm their concordance with one another. Additionally, we compared all sequences to a maternally related reference and verified the identity of the deceased. These results indicate that mtDNA can be successfully extracted from cremated remains and used as a means to establish identity in cases where traditional methods of identification are not feasible. Combination of a calcified tissue-specific DNA extraction method with multiplex PCR and MPS is a useful technique to maximize recovery of genetic information from severely compromised sample types.