CHARACTERIZATION OF UNIDENTIFIED 140-YEAR-OLD HUMAN SKELETAL REMAINS USING MASSIVELY PARALLEL DNA SEQUENCING

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Although the paramount objective of forensic DNA testing of unidentified human remains is positive identification, cases involving historical or archaeological skeletal remains often lack reference samples for comparison. Massively parallel sequencing (MPS) offers an opportunity to provide biometric data in such cases. In this study, MPS was used to provide identity marker information for 140-year-old human skeletal remains from Deadwood, South Dakota, a famous town of the American Old West. Given that the remains were in an unmarked grave and no records existed regarding the identity of the individual, city officials requested the analysis of DNA markers that could help predict the individual's biogeographic ancestry and external physical traits. Results were obtained for 25/26 Y-STRs, 34/34 Y SNPs, 165/165 ancestry-informative SNPs, 28/28 phenotype-informative SNPs, 102/102 human identity SNPs, 27/29 autosomal STRs (plus amelogenin), and 4/8 X-STRs (as well as eleven regions of mtDNA). The Y-chromosome (Y-STR, Y-SNP) and mtDNA profiles of the unidentified skeletal remains are consistent with the R1b and H1 haplogroups, respectively. Both of these haplogroups are the most common haplogroups in Western Europe. Ancestry-informative SNP analysis also supported a European background. The genetic results are consistent with the findings of a previous anthropological report which determined that the remains belong to a male of European ancestry (Caucasian). Phenotype-informative SNP data provided strong support that the individual had light red hair and brown eyes. This study is among the first to genetically characterize historical human remains with forensic genetic marker kits specifically designed for MPS. It demonstrates the potential of MPS to analyze old skeletal samples and to provide substantially more genetic information from the same initial quantities of DNA as that of CE-based analyses.