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Ancient genomics in India: Clarifying the maternal origins of 160-year-old human remains

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Abstract

Sequencing DNA from archaeological remains has opened up new possibilities for furthering our understanding of the origins and evolutionary history of modern humans [1]. However, most ancient DNA (aDNA) studies, thus far, have focused on ancient samples obtained from permafrozen and temperate regions, where preservation conditions are better suited for long-term DNA survival. Consequently, this has left a void in aDNA research in tropical regions such as South Asia. The primary aims of the present study were to (a) test the feasibility of extracting DNA from historical samples (~160 years old) from northern India, and (b) correlate obtained mitochondrial DNA (mtDNA) signatures with geographical origins of the individuals, as reported in historical records. A total of 30 molars were subjected to DNA extractions and Illumina indexed library preparation. All laboratory work was performed following strict aDNA standards in the clean laboratory at the Centre for Cellular and Molecular Biology, Hyderabad. Complete mtDNA genomes were targeted from all 30 samples following the DNA hybridization method outlined in Maricic et al., 2010 [2]. Captured libraries were sequenced on the Illumina HiSeq 2500 platform (100 bp paired-end mode) at MedGenome Inc., Bangalore. Obtained sequences were trimmed for residual adapters using AdapterRemoval and mapped to the revised Cambridge Reference Sequence (rCRS) using BWA. HaploGrep2 [3] was used to assign mtDNA haplogroups to each sample. We successfully obtained endogenous mtDNA sequences from all 30 samples, as confirmed by typical aDNA damage (cytosine deamination on the ends of DNA molecules). Coverage and depth of sequencing were in the range of 91-99.5% and 6X-371X, respectively. To ascertain the maternal origins of the individuals, mtDNA haplogroups of our samples were compared to a database compiled from published mtDNA sequences from modern South Asian individuals. Based on this, we were able to confirm northern Indian origins for the studied individuals, although further fine-scale resolution will only be available with a more comprehensive comparative dataset. This pilot study shows the feasibility of obtaining aDNA from India and its immense value in understanding the population history of humans over time.

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