A PEDIGREE ANALYSIS OF TWO MULTI-GENERATIONAL LINEAGES DETERMINE THE POINT MUTATION RATES OF THE HUMAN MITOCHONDRIAL D-LOOP REGION

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The recent use of mitochondrial deoxyribonucleic acid (mtDNA) analysis for human identification has proven to be a valuable tool when dealing with difficult samples, i.e. skeletal remains, mummified tissues, teeth, and hair.

The isolation of chromosomal DNA from these tissues is often found to be degraded and in quantities too small for analysis. To further complicate matters, the patterns displayed by Mendelian inheritance often make it difficult to determine exact familial relationships when a limited supply of known reference samples are available.

The advantages of mtDNA are that it exhibits a simple non-Mendelian maternal inheritance pattern, has a high copy number per cell, and the hypervariable regions (HVI,HVII) in the D-loop show individual sequence variation with relatively short lengths of base pairs (~300bp). Examples of this methodology are the identification of unknown " war dead" remains, anthropological analysis of Neanderthal skeletal remains, and forensic identification of hair associated with crime scenes.

With the increased use of mtDNA analysis, larger population samples have shown that mutation rates in specific locations of the D-loop region might be greater than first hypothesized.

Specific studies that concentrate on the analysis of point mutation rates are needed to facilitate interpretation of data involving identification of human mtDNA. One possible method would be the use of pedigree analysis in multi-generational lineages with concentration being on identifying point mutations in the hypervariable regions I and II (HVI, HVII). Special attention would be given to the base pair location, the frequency of occurrence, and the type of mutation, within any given family sequence, inferring differences to the general referenced populations.