OBSERVED mtDNA SUBSTITUTIONS AMONG MATERNAL LINEAGES OF THE EUROPEAN ROYALTY

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Mitochondrial DNA profiling is widely used for forensic identification, especially in cases of highly degraded DNA, and where reference samples are often maternal relatives. Due to its non-coding nature, the control region of mtDNA has a higher tolerance for mutations than the surrounding coding regions. Initial studies performed by this laboratory uncovered a control region mutation rate much higher than that predicted by phylogenetic studies, and since then, a steady progression of work has confirmed this initial observation. It is important for forensic interpretation that the rate and pattern of mtDNA mutations between generations be well characterized. We report here a substantial addition to the number of pedigree generations that have been compared for observed mtDNA mutations. The data derive in large part from maternal lineages of the European Royalty, for whom accurate historical records have allowed identification of deep maternal pedigrees. Our comparisons over the entire mtDNA control region span 686 generations and reveal 8 intergenerational substitutions, as well as a greater number of sites with heteroplasmic variants segregating within the lineages. These observations shall be combined with other familial studies to reduce possible stochastic effects, and the observed mutation rate shall be discussed in relation to its possible influence on forensic identification.