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SEQUENCING HUMAN mtDNA TO FIND NEW MUTATIONS LINKED TO MITOCHONDRIAL DISEASE

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Respiration that occurs in mitochondria supplies most of the energy needed for the daily functions needed for cell survival. Thus, a point mutation in the mitochondrial DNA genome could interfere with the proper coding of the specific RNAs and/or protein subunits of the respiratory chain. Depending on how widespread the mutation is, an individual with this type of mutation would then be incapable of generating sufficient energy and would display symptoms of a mitochondrial disease. In this study, approximately 99% of the mitochondrial genome of an individual who displayed these symptoms and who was diagnosed by the Mayo Clinic with a probable mitochondrial disorder was sequenced. In addition to previously reported mutations and polymorphisms, two new point mutations were identified: a silent G-A point mutation located at 12127 nt within the ND4 gene and an A-G point mutation located at 13681 nt, which codes for a T to A amino acid change within the ND5 gene. Also within the ND5 gene and only 27 nt away (13708 nt) from the new mutation, a previously reported G-A mutation was also identified. The combination of these two mutations may affect protein structure and function and could lead to a dysfunction in the ND5 subunit. Knowledge of the presence of mutations and polymorphisms in the mitochondrial DNA genome is important to both medical and forensic communities. Analysis of the heteroplasmic distribution of these mutations from this patient and maternal relatives could prevent some of the ambiquity that arises from sample variation in forensic human identification.