Abstract 34

INVESTIGATION OF TWO DELETIONS FOUND IN THE MITOCHONDRIAL DNA GENOME OF AN INDIVIDUAL SUFFERING FROM SYMPTOMS RELATED TO A MITOCHONDRIAL MYOPATHY

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The mitochondrial genome of a Caucasian female in her mid twenties who had been diagnosed with a mitochondrial myopathy has been sequenced to identify single nucleotide polymorphisms (SNPs) and to seek out deletions within the genome. During the examination of 102 reported Mitomap deletions[1], two unreported deletions were discovered in this individual. Currently, the regions containing the two deletions are being sequenced and further examined to establish the deletion junctions and to determine if these deletions interfere with the coding of functional proteins. It is believed that these deletions may lead to heteroplasmic length polymorphisms within the mtDNA genome and ultimately may contribute to the mitochondrial myopathy symptoms exhibited by the individual. The exploration of deletions and polymorphisms within the human population has important implications for both the medical and forensic communities; this study on the variability of the mitochondrial genome may lead to a greater understanding of the effect of sample variation in the ambiguity often found in the field of forensic human identification.

References

[1] www.mitomap.org