IMPLICATIONS IN THE REPORT OF THE HETEROPLASMIES OF LENGTH IN THE POLY-C TRACT OF MITOCHONDRIAL DNA D-LOOP

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Maternal lineage studies are fundamental in defining familiar links between individuals with the absence of both parents. Approximately 30% of the samples analyzed in our lab present the T16189C mutation that produces length heteroplasmies between 16182 and 16193 positions, which generate a coexistence of multiple variants in the mitochondrial DNA sequence.

This research analyzed 200 samples that have the poli-C tract with the object to study all the variables present and their implication in the use of the ISFG recommended nomenclature.

The HV1 region of every sample was sequenced according to standard protocols using forward and reverse primers. The Sequencing Analysis Software v6.0 and SeqScape Software v3.0 were ran in a 3500 Genetic Analyzer, with a posterior manual reading.

58.5% of the examined samples presented the following mutations: T16189C c16193 16193.1c 16193.2c. Another 26% showed: T16189C 16193.1c 16193.2c. A 4.5%: T16189C 16193.1C 16193.2c 16193.3c. The remaining 11% corresponded to other variables.

According to ISFG current recommendations only the dominant variable is to be included in the report, with no record being left on the rest of them. Given they represent an 85% of the total amount, we consider a thorough report would consist on presenting every variable shown in the samples to provide more accurate information on the analyzed sequence.