Table S8 D-loop variants (complex nucleotide exchanges, insertions and deletions) detected by re-sequencing (Sanger) of mtDNA and frequencies in cases and controls

|  |  |  |
| --- | --- | --- |
| **Detected** **Variants** | **Frequency cases [%]** | **Frequency controls [%]** |
| **n=191** | **n=191** |
| **m.16183A/CC** | 2.62 | 1.05 |
| **m.16183A/CCC a** | 0.52 | 0.00 |
| **m.16189T/CC a** | 1.05 | 2.62 |
| **m.43\_44insG a** | 1.05 | 0.00 |
| **m.297\_298insC a** | 0.00 | 0.52 |
| **m.309\_310insC** | 34.03 | 39.79 |
| **m.309\_310insCC** | 14.14 | 12.57 |
| **m.310\_311insTC** | 0.52 | 1.57 |
| **m.315\_316insC** | 95.29 | 97.91 |
| **m.315\_316insCC** | 1.05 | 0.00 |
| **m.315\_316insCCC** | 0.52 | 0.00 |
| **m.451\_452insT** | 0.52 | 0.52 |
| **m.514\_515insAC** | 5.24 | 8.38 |
| **m.514\_515insACAC** | 3.14 | 1.05 |
| **m.514\_515insACACAC** | 0.52 | 0.52 |
| **m.567\_568insC** | 1.05 | 0.00 |
| **m.567\_568insCCC** | 0.52 | 1.05 |
| **m.567\_568insCCCC** | 0.52 | 0.00 |
| **m.567\_568insCCCCC** | 0.52 | 3.14 |
| **m.576\_577insCA a** | 0.52 | 0.00 |
| **m.16193\_16194insC** | 0.52 | 0.00 |
| **m.310delT** | 1.05 | 0.00 |
| **m.311\_313delCCC** | 0.52 | 0.00 |
| **m.498delC** | 2.62 | 1.05 |
| **m.513\_514delGC a** | 0.00 | 0.52 |
| **m.515\_516delAC** | 10.99 | 10.99 |
| **m.568delC a** | 0.52 | 0.00 |
| **m.16189delT** | 0.52 | 0.00 |
| **m.16257delC** | 0.52 | 0.00 |

a variant has not been described previously based on [www.mitomap.org](file:///C%3A%5CUsers%5CNadja%5CDiss%5Cwww.mitomap.org), last edited on Apr 23, 2013 (Ruiz-Pesini et al. 2007)

Reference:

Ruiz-Pesini E, Lott MT, Procaccio V, Poole JC, Brandon MC, et al. (2007) An enhanced MITOMAP with a global mtDNA mutational phylogeny. Nucleic Acids Res 35 (Database issue):D823-D828.