Supplementary Table 2

the HGVS validation of mtDNA variations mentioned in the study

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **MtDNA variations** | Constructed HGVS variant description | **Genomic description** | **Created** | **Deleted** |
| 249A>d | [NC\_012920.1:m.[249A>d;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B249A%3Ed%3B%3E%5D) | [NC\_012920.1:m.249del](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.249del) |  |  |
| 489T>C | [NC\_012920.1:m.489T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.489T%3EC) | [NC\_012920.1:m.489T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.489T%3EC) |  |  |
| 663A>G | [NC\_012920.1:m.[663A>G;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B663A%3EG%3B%3E%5D) | [NC\_012920.1:m.663A>G](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.663A%3EG) | BstNI, HaeIII, LpnPI (2), PspGI, ScrFI, StyD4I |  |
| 1391T>C | [NC\_012920.1:m.1391T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.1391T%3EC) | [NC\_012920.1:m.1391T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.1391T%3EC) |  |  |
| 3394T>C | [NC\_012920.1:m.[3394T>C;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B3394T%3EC%3B%3E%5D) | [NC\_012920.1:m.3394T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.3394T%3EC) |  |  |
| 4715A>G | [NC\_012920.1:m.4715A>G](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.4715A%3EG) | [NC\_012920.1:m.4715A>G](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.4715A%3EG) | BsrDI, NciI, ScrFI, StyD4I |  |
| 4833A>G | [NC\_012920.1:m.[4833A>G;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B4833A%3EG%3B%3E%5D) | [NC\_012920.1:m.4833A>G](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.4833A%3EG) | BsaHI, HaeII, HhaI, HinP1I, KasI, NarI, PluTI, SfoI |  |
| 5178C>a | [NC\_012920.1:m.5178C>A](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.5178C%3EA) | [NC\_012920.1:m.5178C>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.5178C%3EA) |  | AluI, CviKI\_1 |
| 5417G>A | [NC\_012920.1:m.5417G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.5417G%3EA) | [NC\_012920.1:m.5417G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.5417G%3EA) | MluCI | HpyCH4III |
| 6023G>A | [NC\_012920.1:m.6023G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.6023G%3EA) | [NC\_012920.1:m.6023G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.6023G%3EA) | BmrI, BsrI | AluI, BseYI, NmeAIII |
| 6392T>C | [NC\_012920.1:m.6392T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.6392T%3EC) | [NC\_012920.1:m.6392T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.6392T%3EC) |  |  |
| 8281-8289d | [NC\_012920.1:m.8281\_8289del](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.8281_8289del) | [NC\_012920.1:m.8281\_8289del](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.8281_8289del) |  |  |
| 9824T>C | [NC\_012920.1:m.[9824T>C;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B9824T%3EC%3B%3E%5D) | [NC\_012920.1:m.9824T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.9824T%3EC) | HinfI, MlyI, PleI, XcmI |  |
| 10310G>A | [NC\_012920.1:m.[10310G>A;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B10310G%3EA%3B%3E%5D) | [NC\_012920.1:m.10310G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.10310G%3EA) |  | BfuAI, BspMI, LpnPI |
| 10400C>T | [NC\_012920.1:m.[10400C>T;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B10400C%3ET%3B%3E%5D) | [NC\_012920.1:m.10400C>T](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.10400C%3ET) |  |  |
| 14783T>C | [NC\_012920.1:m.[14783T>C;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B14783T%3EC%3B%3E%5D) | [NC\_012920.1:m.14783T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.14783T%3EC) |  | AseI, PacI |
| 15043G>A | [NC\_012920.1:m.[15043G>A;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B15043G%3EA%3B%3E%5D) | [NC\_012920.1:m.15043G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.15043G%3EA) | Hpy188I |  |
| 16126T>C | [NC\_012920.1:m.[16126T>C;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B16126T%3EC%3B%3E%5D) | [NC\_012920.1:m.16126T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.16126T%3EC) | HpyCH4V |  |
| 16257C>a | [NC\_012920.1:m.[16257C>A;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B16257C%3EA%3B%3E%5D) | [NC\_012920.1:m.16257C>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.16257C%3EA) |  | CviKI\_1 |
| 16261C>T | [NC\_012920.1:m.[16261C>T;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B16261C%3ET%3B%3E%5D) | [NC\_012920.1:m.16261C>T](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.16261C%3ET) |  |  |
| 16274G>A | [NC\_012920.1:m.[16274G>A;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B16274G%3EA%3B%3E%5D) | [NC\_012920.1:m.16274G>A](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.16274G%3EA) |  | BciVI |
| 16311T>C | [NC\_012920.1:m.[16311T>C;>]](https://mutalyzer.nl/name-checker?description=NC_012920.1:m.%5B16311T%3EC%3B%3E%5D) | [NC\_012920.1:m.16311T>C](https://mutalyzer.nl/name-checker?description=NC_012920.1%3Am.16311T%3EC) | MwoI |  |