**Table e-2: Mitochondrial DNA variants associated with the m.15992A>T *MT-TP* mutation**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| ***Position*** | ***P1*** | ***P3*** | ***P4*** | ***P5*** | ***P6*** | ***Ref*** | ***Gene*** | ***AA*** | ***# reports*** |
| 73 | G | G | G | G | G | A | *MT-HV2* |  | 76960 |
| 228 | A | G | G | A | A | G | *MT-HV2* |  | 1870 (A) |
| 263 | G | G | G | G | G | A | *MT-HV2* |  | 86799 |
| 295 | T | T | T | T | T | C | *MT-HV2* |  | 3586 |
| 315 | CC | CC | CC | CC | CC | C | *MT-CSB2* |  | 29290 |
| 462 | T | T | T | T | T | C | *MT-HV3* |  | 2322 |
| 489 | C | C | C | C | C | T | *MT-HV3* |  | 23043 |
| 750 | G | G | G | G | G | A | *MT-RNR1* |  | 48276 |
| 794 | C | C | C | C | C | T | *MT-RNR1* |  | ***37*** |
| 1438 | G | G | G | G | G | A | *MT-RNR1* |  | 46721 |
| 2706 | G | G | G | G | G | A | *MT-RNR2* |  | 38981 |
| 3010 | A | A | A | A | A | G | *MT-RNR2* |  | 7061 |
| 4216 | C | C | C | C | C | T | *MT-ND1* | Y-Y | 4862 |
| 4769 | G | G | G | G | G | A | *MT-ND2* | M-M | 48004 |
| 5024 | T | T | T | T | T | C | *MT-ND2* | T-T | ***15*** |
| 7028 | T | T | T | T | T | C | *MT-CO1* | A-A | 39824 |
| 7098 | T | T | T | T | T | C | *MT-CO1* | L-L | ***5*** |
| 8860 | G | G | G | G | G | A | *MT-ATP6* | **T-A** | 48227 |
| 10398 | G | G | G | G | G | A | *MT-ND3* | **T-A** | 21770 |
| 11251 | G | G | G | G | G | A | *MT-ND4* | L-L | 4574 |
| 11719 | A | A | A | A | A | G | *MT-ND4* | G-G | 38205 |
| 12612 | G | G | G | G | G | A | *MT-ND5* | V-V | 2475 |
| 13708 | A | A | A | A | A | G | *MT-ND5* | **A-T** | 3506 |
| 14766 | T | T | T | T | T | C | *MT-CYB* | **T-I** | 37907 |
| 14798 | C | C | C | C | C | T | *MT-CYB* | **F-L** | 3179 |
| 15326 | G | G | G | G | G | A | *MT-CYB* | **T-A** | 48493 |
| 15452 | A | A | A | A | A | C | *MT-CYB* | **L-I** | 4577 |
| 15992 | T | T | T | T | T | A | *MT-TP* |  | ***1*** |
| 16069 | T | T | T | T | T | C | *MT-HV1* |  | 4663 |
| 16126 | C | C | C | C | C | T | *MT-HV1* |  | 12095 |
| 16168 | T | T | T | T | T | C | *MT-HV1* |  | 1462 |
| 16519 | C | C | C | C | C | T | *Control R* |  | 60910 |

Position=nucleotide position in the reference Cambridge sequence of human mtDNA (GenBank number NC\_012920); P=Patient; Ref=nucleotide observed in the reference Cambridge sequence; AA= consequence on the aminoacid (marked in bold characters when not synonymous); # reports = number of time the variants have been reported in more than 49000 GenBank full length mtDNA sequences and more than 72000 control region mtDNA sequences; very rare variants are indicated in bold italic.