## Table S1

## a.

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Sample ID | Position | Ref | Primary Base | Primary Base Coverage | Secondary Base | Secondary Base Coverage | % Heteroplasmy | Status | 1000 Genomes |
| NA19092 | 150 | C | T | 117 | C | 43 | 27 |   |   |
| NA19203 | 1552 | G | G | 76 | A | 69 | 48 | validated |   |
| NA18500 | 2045 | A | G | 120 | A | 28 | 19 |   |   |
| NA18912 | 2274 | A | A | 81 | G | 46 | 36 | validated | X |
| NA18912 | 2838 | A | A | 85 | G | 24 | 22 |   |   |
| NA19171 | 4227 | A | A | 100 | G | 12 | 11 | validated |   |
| NA19152 | 7364 | A | A | 76 | G | 51 | 40 | validated |   |
| NA18870 | 10567 | T | T | 143 | C | 15 | 9 |   |   |
| NA19200 | 11371 | A | A | 82 | G | 61 | 43 | validated |   |
| NA19140 | 13602 | T | T | 60 | C | 11 | 15 | validated |   |
| NA19137 | 13722 | A | A | 87 | G | 31 | 26 | validated |   |
| NA18870 | 14352 | C | C | 63 | A | 13 | 17 |   | X |
| NA18516 | 14610 | T | T | 62 | C | 14 | 18 |   |   |

**B.**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Sample ID | Position | Ref | Primary Base | Primary Base Coverage | Secondary base | Secondary Base Coverage | % Hetroplasmy | Status | 1000Genomes |
| NA12248 | 73 | A | G | 80 | A | 21 | 21 |   |   |
| NA12248 | 150 | C | T | 100 | C | 22 | 18 |   |   |
| NA12891 | 152 | T | C | 79 | T | 17 | 18 |   |   |
| NA12156 | 195 | T | C | 62 | T | 17 | 22 |   |   |
| NA10856 | 204 | T | C | 87 | T | 27 | 15 |   |   |
| NA12145 | 251 | G | A | 40 | G | 21 | 34 | validated  |   |
| NA11882 | 673 | T | T | 90 | C | 45 | 33 |   |   |
| NA10851 | 1333 | G | G | 92 | A | 67 | 42 | validated\*  |   |
| NA12248 | 1721 | C | T | 115 | C | 14 | 11 |   |   |
| NA12891 | 2259 | C | T | 85 | C | 21 | 20 |   |   |
| NA12145 | 2407 | T | T | 77 | C | 15 | 16 |   |   |
| NA11831 | 2448 | G | G | 75 | A | 21 | 22 |   |   |
| NA12248 | 2706 | A | G | 118 | A | 16 | 12 |   |   |
| NA12891 | 2706 | A | A | 93 | G | 11 | 11 |   |   |
| NA12156 | 2927 | C | C | 116 | T | 19 | 14 | validated  |   |
| NA12248 | 3010 | G | G | 124 | A | 19 | 13 |   |   |
| NA12248 | 3197 | T | C | 115 | T | 16 | 12 |   |   |
| NA12248 | 3212 | C | T | 112 | C | 17 | 13 |   |   |
| NA11882 | 3591 | G | G | 79 | A | 78 | 50 | validated  |   |
| NA12248 | 4067 | T | T | 103 | C | 17 | 14 |   |   |
| Sample ID | Position | Ref | Primary Base | Primary Base Coverage | Secondary base | Secondary Base Coverage | % Hetroplasmy | Status | 1000Genomes |
| NA12760 | 4502 | T | T | 104 | C | 13 | 11 |   |   |
| NA12248 | 4732 | A | G | 89 | A | 11 | 11 |   |   |
| NA12891 | 4745 | A | G | 81 | A | 13 | 14 |   |   |
| NA12248 | 5460 | G | G | 89 | A | 17 | 16 |   |   |
| NA12891 | 6266 | A | A | 50 | G | 21 | 30 |   |   |
| NA12891 | 6755 | G | G | 92 | A | 25 | 21 |   |   |
| NA12145 | 7028 | C | T | 64 | C | 33 | 34 |   |   |
| NA12891 | 7028 | C | C | 95 | T | 20 | 17 |   |   |
| NA12891 | 7337 | G | A | 105 | G | 30 | 22 |   |   |
| NA10863 | 7524 | T | T | 81 | C | 25 | 24 |   |   |
| NA10863 | 7925 | G | G | 82 | A | 46 | 36 | validated  |   |
| NA12156 | 8512 | A | A | 114 | G | 21 | 16 | validated  |   |
| NA12145 | 9055 | G | A | 39 | G | 35 | 47 |   |   |
| NA12146 | 9095 | T | C | 45 | T | 43 | 49 | validated  |   |
| NA12156 | 9501 | T | T | 96 | C | 16 | 14 |   |   |
| NA12145 | 9698 | T | C | 63 | T | 37 | 37 |   |   |
| NA12145 | 10398 | A | G | 58 | A | 39 | 40 |   |   |
| NA12145 | 10550 | A | G | 57 | A | 47 | 45 |   |   |
| NA12145 | 11299 | T | C | 44 | T | 26 | 37 |   |   |
| NA12145 | 11467 | A | A | 44 | G | 39 | 47 |   |   |
| NA12144 | 12303 | C | C | 134 | T | 25 | 16 |   | X |
| NA12248 | 12372 | G | A | 80 | G | 12 | 13 |   |   |
| NA11882 | 12642 | A | A | 118 | G | 53 | 31 |   |   |
| NA07019 | 13328 | C | C | 70 | T | 10 | 13 | validated  |   |
| NA10854 | 13466 | G | G | 152 | A | 26 | 15 |   |   |
| NA12760 | 13558 | G | A | 73 | G | 36 | 33 |   |   |
| NA12248 | 13617 | T | C | 89 | T | 16 | 15 |   |   |
| NA12248 | 13637 | A | G | 84 | A | 15 | 15 |   |   |
| NA12891 | 14872 | C | T | 107 | C | 13 | 11 |   |   |
| NA12248 | 14902 | C | C | 85 | T | 10 | 11 |   |   |
| NA12248 | 14956 | T | C | 91 | T | 11 | 11 | validated  |   |
| NA11995 | 15663 | T | T | 191 | C | 23 | 11 |   |   |
| NA12248 | 16172 | T | T | 60 | C | 10 | 14 |   |   |
| NA12248 | 16192 | C | C | 57 | T | 8 | 12 |   |   |
| NA12248 | 16270 | C | T | 53 | C | 10 | 16 |   |   |
| NA12145 | 16284 | A | A | 55 | G | 8 | 13 | validated  |   |
| NA12248 | 16312 | A | A | 59 | G | 11 | 16 |   |   |
| NA12248 | 16519 | T | T | 90 | C | 15 | 14 |   |   |
| \*validated by TaqMan as well |  |  |  |  |  |  |