

Table 1 | Variants discovered by project, type, population and novelty

a Summary of project data including combined exon populations

| Statistic | Low coverage | | | | Trios | | | Exon (total) | Union across projects |
|--|--------------------|---------------------|--------------------|---------------------|--------------------|--------------------|--------------------|-----------------|-----------------------|
| | CEU | YRI | CHB+JPT | Total | CEU | YRI | Total | | |
| Samples | 60 | 59 | 60 | 179 | 3 | 3 | 6 | 697 | 742 |
| Total raw bases (Gb) | 1,402 | 874 | 596 | 2,872 | 560 | 615 | 1,175 | 845 | 4,892 |
| Total mapped bases (Gb) | 817 | 596 | 468 | 1,881 | 369 | 342 | 711 | 56 | 2,648 |
| Mean mapped depth (×) | 4.62 | 3.42 | 2.65 | 3.56 | 43.14 | 40.05 | 41.60 | 55.92 | NA |
| Bases accessed (% of genome) | 2.43 Gb (86%) | 2.39 Gb (85%) | 2.41 Gb (85%) | 2.42 Gb (86.0%) | 2.26 Gb (79%) | 2.21 Gb (78%) | 2.24 Gb (79%) | 1.4 Mb | NA |
| No. of SNPs (% novel) | 7,943,827 (33%) | 10,938,130 (47%) | 6,273,441 (28%) | 14,894,361 (54%) | 3,646,764 (11%) | 4,502,439 (23%) | 5,907,699 (24%) | 12,758 (70%) | 15,275,256 (55%) |
| Mean variant SNP sites per individual | 2,918,623 | 3,335,795 | 2,810,573 | 3,019,909 | 2,741,276 | 3,261,036 | 3,001,156 | 763 | NA |
| No. of indels (% novel) | 728,075 (39%) | 941,567 (52%) | 666,639 (39%) | 1,330,158 (57%) | 411,611 (25%) | 502,462 (37%) | 682,148 (38%) | 96 (74%) | 1,480,877 (57%) |
| Mean variant indel sites per individual | 354,767 | 383,200 | 347,400 | 361,669 | 322,078 | 382,869 | 352,474 | 3 | NA |
| No. of deletions (% novel) | ND | ND | ND | 15,893 (60%) | 6,593 (41%) | 8,129 (50%) | 11,248 (51%) | ND | 22,025 (61%) |
| No. of genotyped deletions (% novel) | ND | ND | ND | 10,742 (57%) | ND | ND | 6,317 (48%) | ND | 13,826 (58%) |
| No. of duplications (% novel) | 259 (90%) | 320 (90%) | 280 (91%) | 407 (89%) | 187 (93%) | 192 (91%) | 256 (92%) | ND | 501 (89%) |
| No. of mobile element insertions (% novel) | 3,202 (79%) | 3,105 (84%) | 1,952 (76%) | 4,775 (86%) | 1,397 (68%) | 1,846 (78%) | 2,531 (78%) | ND | 5,370 (87%) |
| No. of novel sequence insertions (% novel) | ND | ND | ND | ND | 111 (96%) | 66 (86%) | 174 (93%) | ND | 174 (93%) |

b Exon populations separately

| Statistic | CEU | TSI | LWK | YRI | CHB | CHD | JPT |
|------------------------------------|-------------|-------------|-------------|-------------|-------------|-------------|-------------|
| Samples | 90 | 66 | 108 | 112 | 109 | 107 | 105 |
| Total collected bases (Gb) | 151 | 64 | 53 | 147 | 93 | 127 | 211 |
| Mean mapped depth on target (×) | 73 | 71 | 32 | 62 | 47 | 62 | 53 |
| No. of SNPs (% novel) | 3,489 (34%) | 3,281 (34%) | 5,459 (50%) | 5,175 (46%) | 3,415 (47%) | 3,431 (50%) | 2,900 (42%) |
| Variant SNP sites per individual | 715 | 727 | 902 | 794 | 713 | 770 | 694 |
| No. of indels (no. novel) | 23 (10) | 22 (11) | 24 (16) | 38 (21) | 30 (16) | 26 (13) | 25 (11) |
| Variant indel sites per individual | 3 | 3 | 3 | 3 | 3 | 2 | 3 |

NA, not applicable; ND, not determined.