

Supplementary materials

Supplementary Table 1 RNA sequencing coverage and quality statistics per sample

| Sample ID | Total number of sequenced reads | Total number of mapped reads <sup>a</sup> | Ratio of uniquely mapped reads to rRNA (rRNA rate) | Ratio of all reads aligned to total reads mapped (expression efficiency) | Total number of exon-mapped reads uniquely detected transcripts with profile reads $\geq 1$ <sup>b</sup> |
|-----------|---------------------------------|-------------------------------------------|----------------------------------------------------|--------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------|
| A1        | 196.02                          | 163.01                                    | 10.88                                              | 0.58                                                                     | 14286                                                                                                    |
| A4        | 160.18                          | 108.70                                    | 22.64                                              | 0.56                                                                     | 13254                                                                                                    |
| A5        | 238.51                          | 208.79                                    | 5.01                                               | 0.62                                                                     | 13163                                                                                                    |
| A7        | 176.86                          | 126.77                                    | 10.01                                              | 0.56                                                                     | 13388                                                                                                    |
| A6        | 142.37                          | 128.46                                    | 3.2                                                | 0.64                                                                     | 13252                                                                                                    |
| A9        | 156.37                          | 37.36                                     | 60.34                                              | 0.29                                                                     | 13521                                                                                                    |
| A3        | 106.29                          | 97.78                                     | 0.72                                               | 0.67                                                                     | 13518                                                                                                    |
| A11       | 93.02                           | 79.33                                     | 6.07                                               | 0.58                                                                     | 12588                                                                                                    |
| A8        | 79.46                           | 66.08                                     | 14                                                 | 0.55                                                                     | 12549                                                                                                    |
| A14       | 113.48                          | 101.24                                    | 1.91                                               | 0.65                                                                     | 12400                                                                                                    |
| A12       | 53.46                           | 45.04                                     | 13.12                                              | 0.59                                                                     | 13437                                                                                                    |
| A13       | 152.05                          | 136.83                                    | 5.88                                               | 0.65                                                                     | 12131                                                                                                    |
| A27       | 78.97                           | 10.28                                     | 26.23                                              | 0.29                                                                     | 13623                                                                                                    |
| A16       | 58.1                            | 51.71                                     | 3.82                                               | 0.62                                                                     | 13550                                                                                                    |
| A21       | 80.07                           | 42.75                                     | 39.92                                              | 0.3                                                                      | 12444                                                                                                    |

|     |        |        |       |      |       |
|-----|--------|--------|-------|------|-------|
| A18 | 83.34  | 65.76  | 9.87  | 0.55 | 13522 |
| A19 | 55.2   | 48.34  | 4.05  | 0.63 | 12994 |
| A17 | 64.15  | 56.11  | 5.3   | 0.62 | 12407 |
| A28 | 81.07  | 61.87  | 21.28 | 0.39 | 13661 |
| A29 | 72.78  | 31.55  | 54.8  | 0.21 | 13558 |
| A30 | 87.87  | 78.23  | 0.54  | 0.6  | 12786 |
| A31 | 74.88  | 43.65  | 42.61 | 0.25 | 13317 |
| A32 | 88.02  | 81.15  | 0.14  | 0.65 | 13025 |
| A33 | 94.62  | 66.87  | 15.62 | 0.41 | 13526 |
| A34 | 116.76 | 105.82 | 0.24  | 0.62 | 12813 |
| A35 | 105.99 | 96.26  | 0.3   | 0.68 | 12820 |
| A38 | 85.88  | 77.76  | 0.2   | 0.63 | 13249 |
| A36 | 128.93 | 119.58 | 0.3   | 0.65 | 13579 |
| A37 | 87.52  | 80.68  | 0.25  | 0.67 | 11885 |
| A39 | 112.48 | 102.78 | 0.11  | 0.65 | 13144 |
| A26 | 79.62  | 48.82  | 8.36  | 0.34 | 12325 |
| A25 | 87.26  | 55.70  | 8.49  | 0.47 | 13445 |
| A22 | 79.78  | 65.16  | 8.63  | 0.59 | 12929 |

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<sup>a</sup>The reference genome used for alignment and analysis was hg19.

<sup>b</sup>A higher minimum coverage threshold is permitted.

**Supplementary Table 2 Whole-exome sequencing coverage and quality statistics per sample**

| Sample ID | Total number of sequenced reads | Total number of mapped reads <sup>a</sup> | Total number of uniquely non-duplicate covered bases <sup>b</sup> | Median coverage (and targeted range) per base <sup>b</sup> | Percentage of bases with coverage ≥ 10 <sup>b,c</sup> |
|-----------|---------------------------------|-------------------------------------------|-------------------------------------------------------------------|------------------------------------------------------------|-------------------------------------------------------|
| A1        | 44.46                           | 44.42                                     | 2.11                                                              | 1091                                                       | 15.79                                                 |
| A4        | 31.04                           | 31                                        | 2.26                                                              | 1051                                                       | 24.32                                                 |
| A3        | 64.16                           | 53                                        | 227.75                                                            | 172.97                                                     | 86.92                                                 |
| A7        | 159.11                          | 53                                        | 207.17                                                            | 340.11                                                     | 89                                                    |
| A11       | 176.42                          | 49                                        | 230.64                                                            | 322.15                                                     | 87.16                                                 |
| A14       | 146.85                          | 50.5                                      | 201.62                                                            | 294.56                                                     | 88.02                                                 |
| A12       | 179.58                          | 51                                        | 222.01                                                            | 335.59                                                     | 85.86                                                 |
| A13       | 156.83                          | 50                                        | 213.59                                                            | 307.75                                                     | 87.1                                                  |
| A27       | 58.83                           | 53                                        | 168.84                                                            | 115.19                                                     | 88.79                                                 |
| A9        | 93.45                           | 53                                        | 167.92                                                            | 144.68                                                     | 87.89                                                 |
| A8        | 43.74                           | 53                                        | 162.17                                                            | 60.11                                                      | 88.57                                                 |
| A6        | 51.7                            | 53                                        | 166.68                                                            | 84.92                                                      | 88.81                                                 |
| A5        | 91.41                           | 52                                        | 173.93                                                            | 147.92                                                     | 88.77                                                 |
| A35       | 121.22                          | 51                                        | 185.2                                                             | 137.67                                                     | 59.25                                                 |
| A16       | 155.47                          | 49                                        | 221.5                                                             | 357.97                                                     | 86.56                                                 |
| A21       | 156.85                          | 52                                        | 215.15                                                            | 335.54                                                     | 83.05                                                 |

|     |        |      |        |        |       |
|-----|--------|------|--------|--------|-------|
| A22 | 144.96 | 51.5 | 222.17 | 322.65 | 83    |
| A18 | 110.99 | 50   | 193.2  | 260.39 | 88.74 |
| A19 | 98.34  | 48   | 223.79 | 235.81 | 86.41 |
| A17 | 116.12 | 50   | 191.81 | 253.84 | 88.08 |
| A25 | 136.56 | 53   | 213.66 | 277.28 | 83.25 |
| A26 | 94.4   | 56   | 165.85 | 193.38 | 83.68 |
| A28 | 146.25 | 52   | 222.76 | 334.77 | 85.68 |
| A29 | 161.68 | 52   | 219.91 | 351.72 | 85.59 |
| A30 | 138.19 | 52   | 215.66 | 301.71 | 86.23 |
| A31 | 133.48 | 51   | 213.1  | 286.49 | 84.33 |
| A32 | 200.33 | 52   | 212.12 | 397.24 | 85.11 |
| A33 | 122.4  | 52   | 185.48 | 154.78 | 61.73 |
| A34 | 113.4  | 52   | 231.68 | 272.04 | 85.3  |
| A38 | 100.65 | 52   | 223.13 | 215.4  | 84.06 |
| A36 | 151.15 | 52   | 234.11 | 309.2  | 84.8  |
| A37 | 133.28 | 52   | 212.91 | 276.81 | 85.09 |
| A39 | 138.98 | 53   | 215.37 | 275.77 | 84.91 |

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<sup>a</sup>The reference genome used for alignment and analysis was hg19.

<sup>b</sup>After removing unmapped, non-uniquely mapped and duplicate reads.

<sup>c</sup>targeted bases refer to the whole exome.