## SUPPLEMENTARY FIGURE LEGENDS

Supplementary Fig. 1. Karyotypes of the three subjects: (a) jg1a, (b) jg1b, and (c) jg1c. The arrow in panel (a) indicates the normal variation inv(9)(p12q13).

Supplementary Fig. 2. Harr plot of the alignment between chromosome 9 of GRCh38 and two largest scaffolds aligned to chromosome 9 from the (a) jg1a, (b) jg1b, and (c) jg1c assemblies, indicating that the two individual genomes harbor a possible shared inversion. 'Super-scaffold' is the default prefix designated by BionanoSolve software.

Supplementary Fig. 3. Workflow of the construction of JG1. (a) Workflow of the construction of each draft assembly. (b) Workflow of the integration of three draft assemblies. Rectangles indicate substrates such as reads, contigs, and scaffolds. Rectangles with rounded corners indicate software or processes.

Supplementary Fig. 4: Histogram of PacBio subreads length for (a) jg1a, (b) jg1b, and (c) jg1c. The length of each subread was calculated using the SAMtools (ver. 1.8) faidx command.

Supplementary Fig. 5: Histogram of Bionano data for (a) Nt.BspQI of jg1a, (b) Nb .BssSI of jg 1 a, (c) jg 1 b , and (d) jg 1 c . The length of each molecule was extracted from the BNX file.

Supplementary Fig. 6: Majority decision. (a) Schematic representation of majority decision approach. (b) Venn diagram of SNVs detected in JG1, jg1a, jg1b, and jg1c by comparison with hs37d5. The intersection relationship was inferred using the BCFtools (ver. 1.8) isec command.

Supplementary Fig. 7: Length distributions of detected transposable elements in the GRCh38 and JG1 genomes. Shown are Alu, SVA, and LINE1. Transposable elements and their subclasses were identified using RepeatMasker software (ver. 4.0.7) with the 'species human' option. The resulting OUT format files were converted to BED format using the rmsk2bed command of BEDOPS software ${ }^{59}$ (ver. 2.4.35). Transposable elements disrupted by other elements were counted as distinct.

b


C


Supplementary Fig. 1; Takayama et al.


Supplementary Fig. 2
Takayama et al.


Supplementary Fig. 3; Takayama et al


Supplementary Fig. 4
Takayama et al.


Supplementary Fig. 5
Takayama et al.


Supplementary Fig. 6
Takayama et al.


Supplementary Table 1. Basic statistics of PacBio subreads.

| Individual | Number of subreads | Sum of subread length (bp) | depth* |
| :---: | ---: | ---: | ---: |
| jg1a | $34,445,474$ | $364,777,563,591$ | 122 X |
| jg1b | $36,798,731$ | $370,437,373,175$ | 123 X |
| jg1c | $41,535,337$ | $383,220,406,482$ | 128 X |

* Depth is calculated by assuming the genome size $=3.0 \mathrm{~Gb}$.

Supplementary Table 2. Basic statistics of Bionano molecules.

| Individual | Enzyme | Number of <br> molecules | Sum of molecule length <br> (bp) | depth* |
| :---: | :---: | ---: | ---: | ---: |
| jg1a | BspQI | $1,156,682$ | $368,075,072,000$ | 123 X |
|  | BssSI | $1,834,771$ | $418,513,858,000$ | 140 X |
| jg1b | DLE-1 | $2,840,733$ | $480,476,071,000$ | 160 X |
| jg1c | DLE-1 | $3,594,225$ | $524,851,027,000$ | 175 X |

* Depth is calculated by assuming the genome size $=3.0 \mathrm{~Gb}$.

Supplementary Table 3. Basic statistics of Illumina paired-end and mate-pair reads.

| Method | Individual | read length <br> (bp) | \# of reads | Sum of read length (bp) | depth* |
| :---: | :---: | :---: | :---: | :---: | :---: |
| paired end | jg1a | 162 | 543,599,992 | 88,063,198,704 | 29X |
|  |  | 259 | 303,625,608 | 78,639,032,472 | 26X |
|  | jg1b | 162 | 578,161,124 | 93,662,102,088 | 31X |
|  |  | 259 | 319,177,020 | 82,666,848,180 | 28X |
|  | jg1c | 162 | 571,414,220 | 92,569,103,640 | 31X |
|  |  | 259 | 302,332,088 | 78,304,010,792 | 26X |
| mate pair** | jg1a | 201 | 189,189,310 | 38,027,051,310 | 13X |
|  | jg1b |  | 184,346,446 | 37,053,635,646 | 12X |
|  | jg1c |  | 185,928,504 | 37,371,629,304 | 12X |

[^0]Supplementary Table 4. Basic statistics of Bionano assembly.

| Individual | Enzyme | \# of fragments | N50 (Mb) | Total length (Mb) |
| :---: | :---: | ---: | ---: | ---: |
| jg1a | BspQI | 4,761 | 1.179 | 3846.912 |
| jg1a | BssSI | 4,392 | 1.034 | 3202.036 |
| jg1b | DLE-1 | 581 | 41.761 | 3194.487 |
| jg1c | DLE-1 | 496 | 64.293 | 3481.086 |

Supplementary Table 5. Length of consecutive Ns inserted manually.

| chr | pter (bp) | cen (bp) | qter (bp) | References |
| :---: | :---: | :---: | :---: | :---: |
| 1 | 10,000 | 30,000,000 | 10,000 | 48-50 |
| 2 | 10,000 | 3,000,000 | 10,000 |  |
| 3 | 10,000 | 3,000,000 | 10,000 |  |
| 4 | 10,000 | 3,000,000 | 10,000 |  |
| 5 | 10,000 | 3,000,000 | 10,000 |  |
| 6 | 10,000 | 3,000,000 | 10,000 |  |
| 7 | 10,000 | 3,000,000 | 10,000 |  |
| 8 | 10,000 | - | 10,000 |  |
| 9 | 10,000 | 30,000,000 | 10,000 | 48-50 |
| 10 | 10,000 | 3,000,000 | 10,000 |  |
| 11 | 10,000 | - | 10,000 |  |
| 12 | 10,000 | 3,000,000 | 10,000 |  |
| 13 | 16,000,000 | - | 10,000 | 50 |
| 14 | 16,000,000 | - | 10,000 | 50 |
| 15 | 17,000,000 | - | 10,000 | 50 |
| 16 | 10,000 | 20,000,000 | 10,000 | 48-50 |
| 17 | 10,000 | 3,000,000 | 10,000 |  |
| 18 | 10,000 | 3,000,000 | 10,000 |  |
| 19 | 10,000 | 3,000,000 | 10,000 |  |
| 20 | 10,000 | 3,000,000 | 10,000 |  |
| 21 | 11,000,000 | - | 10,000 |  |
| 22 | 13,000,000 | - | 10,000 |  |
| X | 10,000 | 3,000,000 | 10,000 |  |
| Y | 2,260,577 | 3,000,000 | 30,000,000 | 48, 51-53 |


[^0]:    * Depth is calculated by assuming the genome size $=3.0 \mathrm{~Gb}$.
    **all reads (before library separation)

