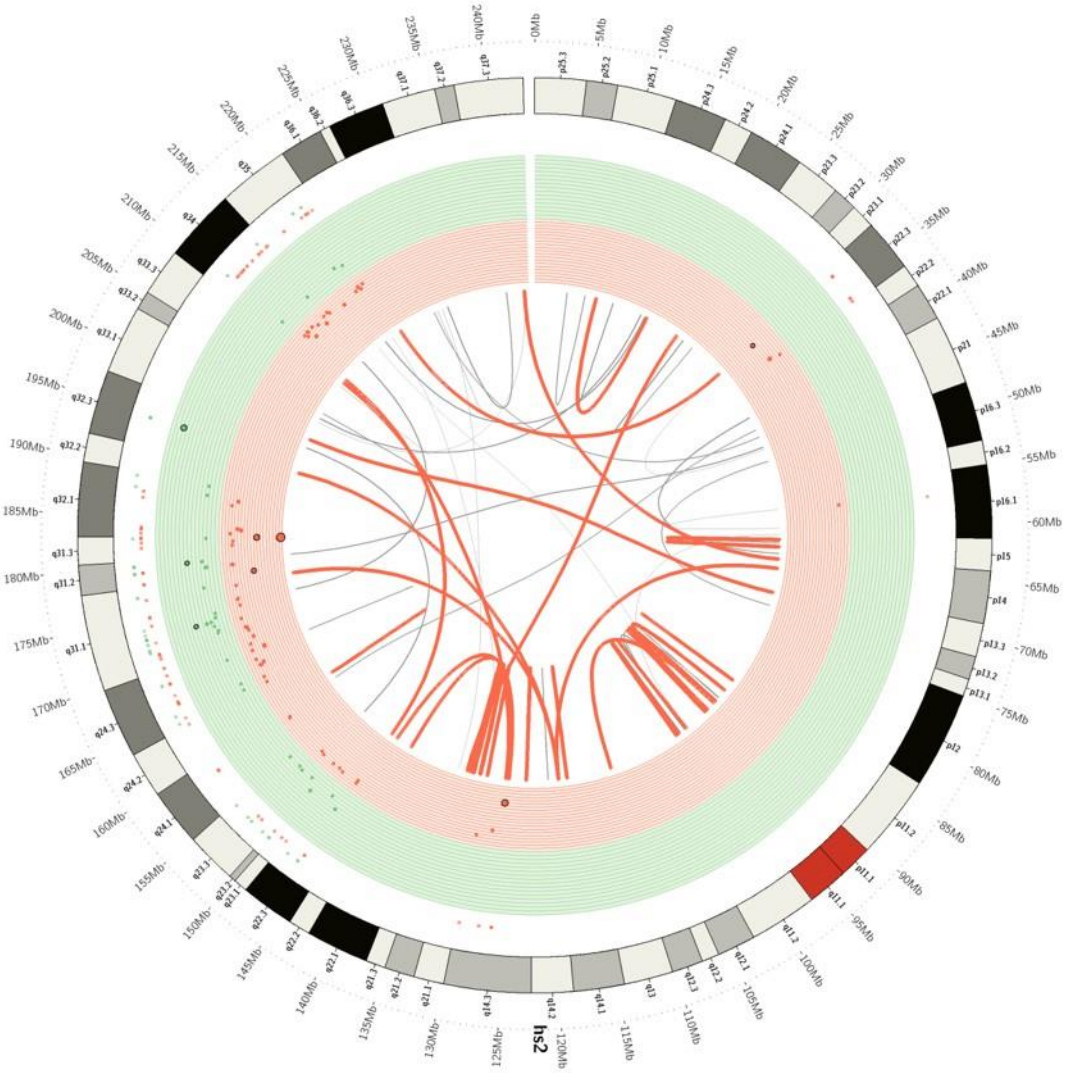
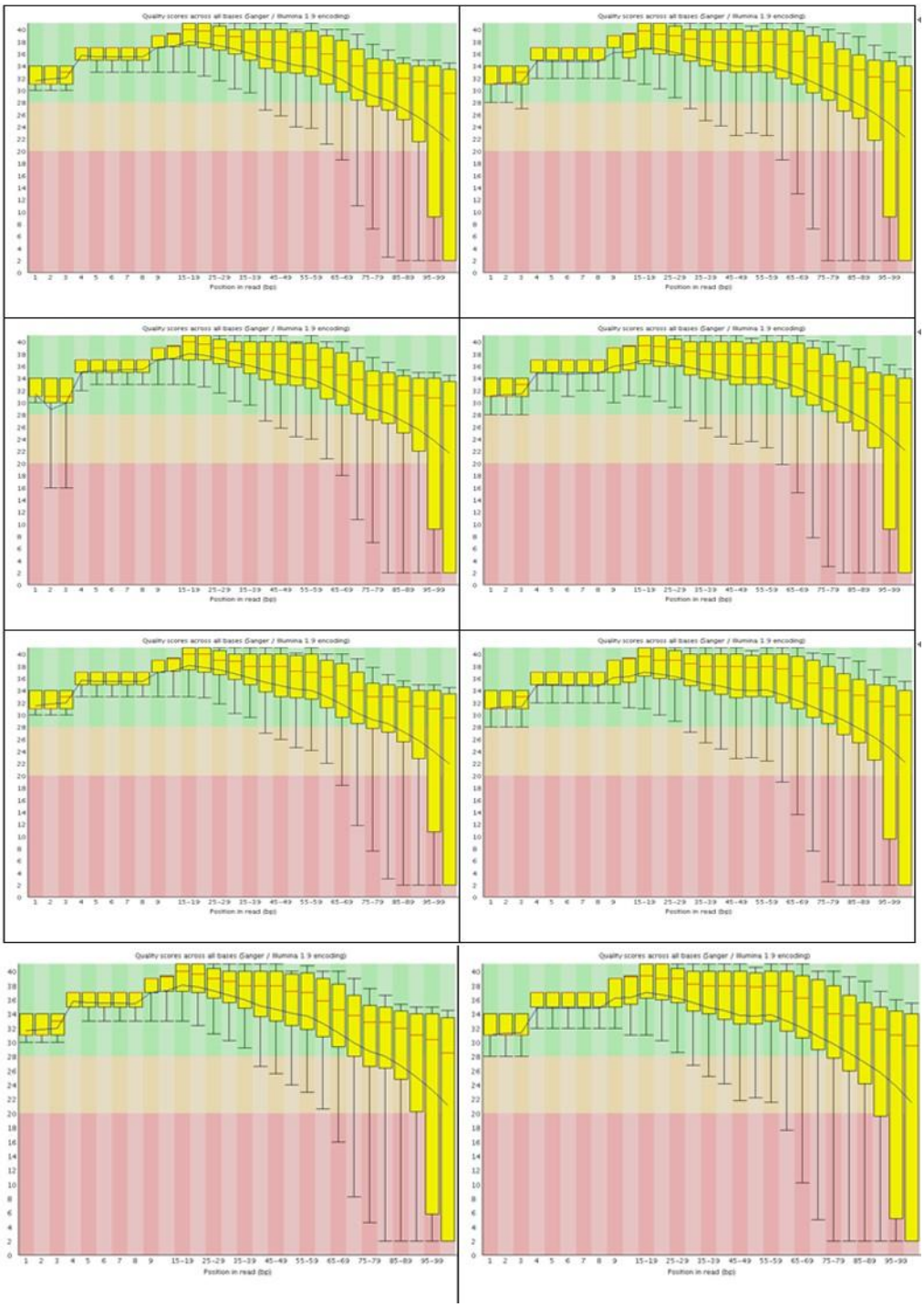
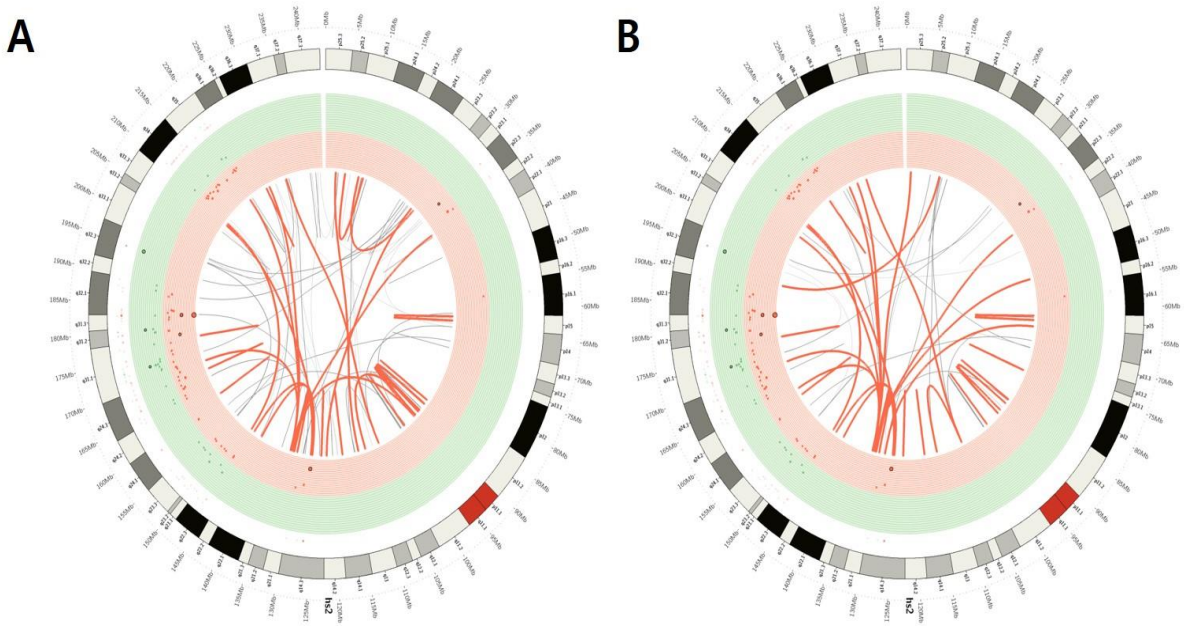
Supplementary figure 1S. Circos plot of inversions on chromosome 2 detected by whole genome sequence data. Outer green dots and inner red dots represent copy number amplification and deletion in the array comparative genomic hybridization **(**CGH), respectively. Red curves are plotted to point both boundaries of each inversion with a high confidence score (>80) and grey curves are plotted to point to those with lower confidence scores.



Supplementary figure 2S. Quality per base of sequence reads. Quality per base of sequence reads decreased while the length of the reads increased and the 10th percentile of the average quality score at the 60th base pair position was lower than 20.



Supplementary figure 3S. Circos plot of inversions on chromosome 2 detected by trimmed reads. Trimmed reads were mapped to (A) GRCh37 and (B) Korean (KSJ) human reference genomes. Outer green dots and inner red dots represent copy number amplification and deletion in the array CGH, respectively. Red curves are plotted to point to both boundaries of each inversion with high confidence scores (>80) and grey curves are plotted to point those with lower confidence scores.



Supplementary figure 4S. Schematic overview of genomic position of the analyzed inversion and neighboring genes. The genomic positions of the 5' and 3' breakpoints are given before (upper) and after (Lower) inversion, and the sizes (Mb) of the fragments involved are indicated. The whole genome analysis validated the breakpoint of inversion to genomic location at 126,101,173 (2q14) and 209,310,827 (2q34) causing separation of the *PTH2R* gene at intron 7. The 2q14 breakpoint is about 430kb and 1Mb away from the nearest transcriptional units, and those nearby transcriptional units (*CNTNAP5* and *GYPC*) are not be disrupted by the inversion. Consequently, the *PTH2R* gene is thought to be fused with the intergenic sequence between *CNTNAP5* and *GYPC*.