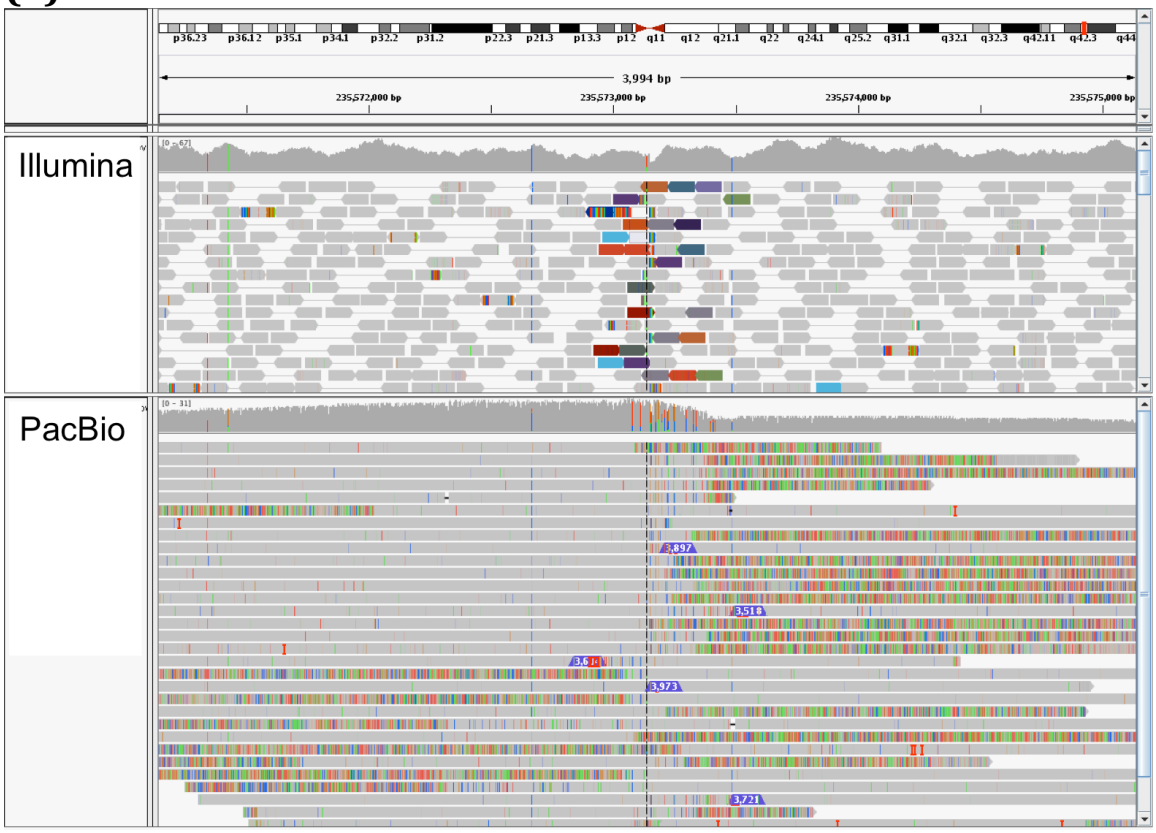
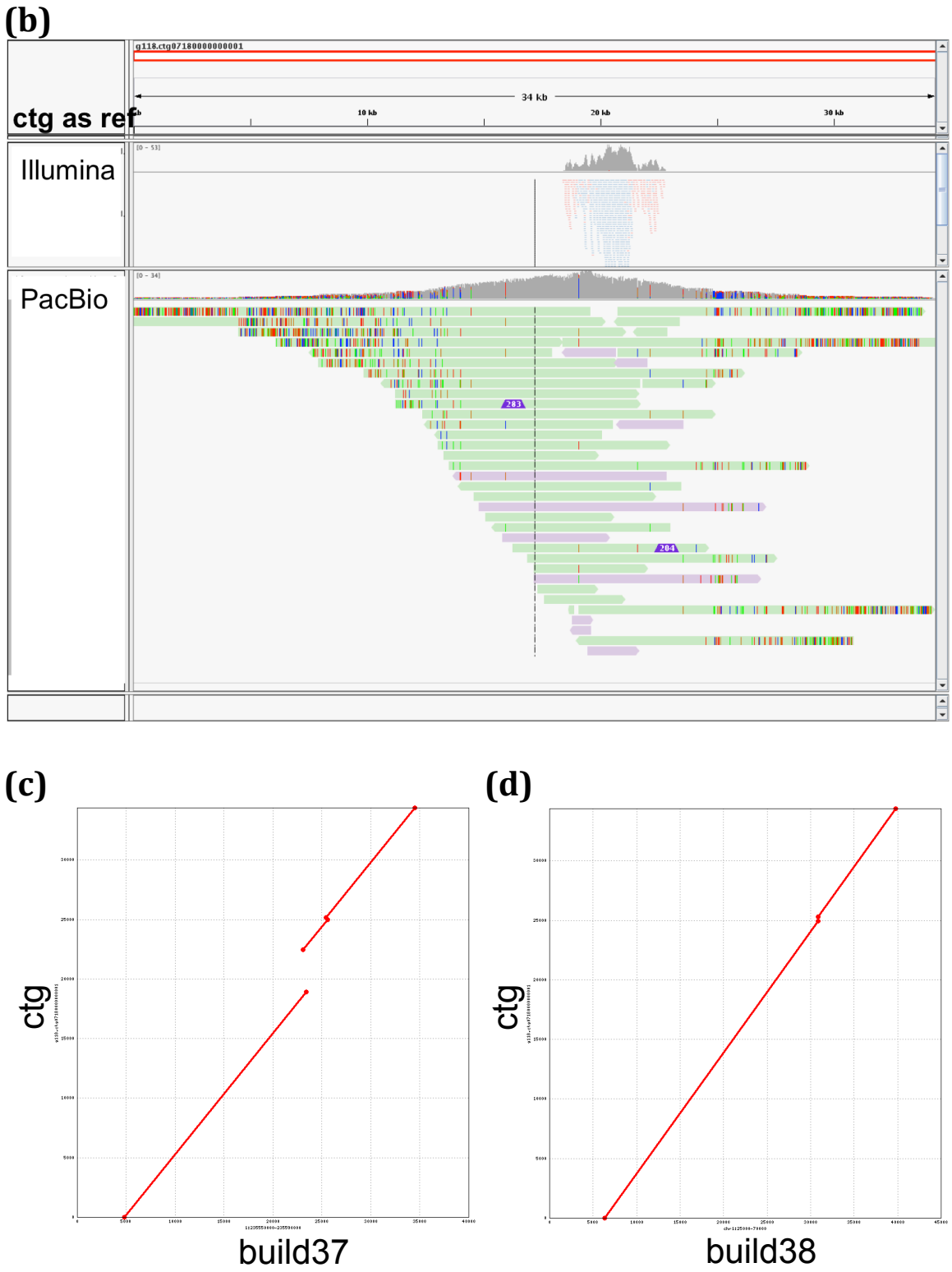
(a)


p36.23 p36.12 p35.1 p34.1 p32.2 p31.2 p22.3 p21.3 p13.3 p12 q11 q12 q21.1 q22 q24.1 q25.2 q31.1 q32.1 q32.3 q42.11 q42.3 q44
3,994 bp
235,572,000 bp
235,573,000 bp
235,574,000 bp
235,575,000 bp
Illumina
[0 - 67]
PacBio
[0 - 31]
3,897
3,518
3,973
3,721



Supplemental Fig. 2 A 3.84kbp novel insertion at 1:235573140 validated by NCBI build v38. (a) Screenshot of Integrative Genomics Viewer (IGV) of reference alignment of the focal region 1:235571140-235575140. The upper panel shows

Illumina reads' alignment; the bottom panel shows PacBio reads' alignment. The IGV is a version with improved PacBio support (consensus mode, secondary alignments filtered), downloaded from https://github.com/amwenger/igv/tree/amw-pb-consensus-mode (same for b). (b) IGV screenshot of the alignment of extracted Illumina (upper panel) and PacBio (bottom panel) reads to the assembled contig. The reads are grouped in color by alignments. For Illumina reads' alignment, blue bars represent the reads whose both ends were unmapped to the reference; red bars represent the reads which are soft clipped, or aligned discordantly. For PacBio reads' alignment, purple bars represent the reads that never appeared in the focal region in reference alignment; green bars represent the reads that have alignments to the focal region. (c) Dot plot of the alignment of the contig to 1:235550000-235590000 on NCBI build v37 using nucmer (Delcher et al. 1999). (d) Dot plot of the alignment of the contig to Homo sapiens chromosome 1 genomic patch of type NOVEL, GRCh38.p7 PATCHES HSCHR1_5_CTG32_1 (25000-70000) using nucmer.

Reference

Delcher AL, Kasif S, Fleischmann RD, Peterson J, White O, Salzberg SL. 1999. Alignment of whole genomes. *Nucleic Acids Res* **27**: 2369-2376.