

Figure S1. Examples of discordant variants between TSO500 and OCP visualized through integrative genomic viewer. The upper half of each panel represents aligned reads of TSO500, and the lower half represents aligned reads of OCP. (A) Variants beyond the OCP targeted regions. There was a ERBB3 c.1463G>A (p.R488Q) variant detected by TSO500. In OCP, no read was present in this region and therefore no variant was reported by OCP. (B) Variants in homopolymer region. There was a PTEN c.V290Sfs*8 (p.867dupA) variant clearly observed in the aligned reads of TSO500 and OCP. TSO500 accurately reported this variant, whereas OCP missed this variant and therefore did not report it. (C) Likely artifactual variants in homopolymer region. In the aligned reads of OCP, some reads showed an insertion of a single A nucleotide, and some showed an insertion of double A nucleotides. Due to these alterations, OCP reported that there was a BRCA2 c.2916_2917insA (p.S973fs) variant in this region. However, in the aligned reads of TSO500, no variant can be observed. As false indels in homopolymer repeats are a well-known weakness of the Ion Torrent sequencing platform, which is used in OCP, BRCA2 c.2916_2917insA (p.S973fs) is likely to be an artifactual variant in homopolymer region introduced by errors of sequencing. (D) Inappropriately annotated variant due to misalignment. In the aligned reads of TSO500, a three-nucleotide deletion of TP53 c.419_421delCCT (p.T140_C141delinsS) can be clearly observed. The alignment of OCP in this region is more disorganized. Although the three-nucleotide deletion of c.419_421delCCT still can be seen, OCP erroneously reported this variant as TP53 c.421T>A (p.C141S). TSO500, TruSight Oncology 500; OCP, Oncomine Comprehensive Assay Panel v3.

