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GIGA-D-17-00028
"Alignment of 1000 Genomes Project Reads to Reference Assembly GRCh38"
Original Submission

Valerie Schneider (Reviewer 2)

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This is a clear, well-written manuscript describing the realignment of read data from the 1000 Genomes Project to an analysis set including GRCh38, an updated version of the human reference genome assembly. This mapping task is a necessary step in the larger task of re-calling variants on the newer assembly, which the authors cite as their intention. The realignment detailed here will provide for more robust variant calling on GRCh38 and better data interpretation than variant remapping, which is compromised in regions of the assembly that have undergone change and no longer align well between versions. I support the publication of this manuscript, provided the following minor criticisms are addressed:

1. p. 3, line 35 (Bullet point #) "Substantially increase the number of alternative haplotypes associated with the assembly"
The word "haplotypes" should be modified, as the Genome Reference Consortium did not require that the alternate loci scaffolds be haplotype-specific, though it is true that they increase sequence diversity in the reference (see Box 1, https://www.ncbi.nlm.nih.gov/pubmed/21750661).

2. p. 4, line 57 "the Epstein-Barr virus (EBV)sequence, ALT contigs, and the decoy sequences."
Provide the INSDC accessions for EBV (AJ507799.2) and the decoy (GCA_000786075.2).

3. p. 4, line 56-58 "...but they are included in the reference"
The word "reference" is unclear, as it may be interpreted to mean "reference assembly", rather than the alignment target reference. Suggestion: replace "reference" with "analysis set" or similar phrase.

4. p. 5, line 6 "The reference data set..."
Similarly, a suggestion to replace "reference" with "analysis".

5. p. 6, line
Provide a URL for the NCBI Remapper. Suggested: https://www.ncbi.nlm.nih.gov/genome/tools/remap.

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