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Tool Updates

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haplotypecaller:

- Adds the following new options:
 - --minimum-mapping-quality
 - --mapping-quality-threshold-for-genotyping
 - --enable-dynamic-read-disqualification-for-genotyping
- Improved performance by leveraging AVX512 instructions for CPU-based PairHMM computation.

mutectcaller:

• Improved performance by leveraging AVX512 instructions for CPU-based PairHMM computation.

deepvariant:

- Updates to match the baseline version v1.6.1.
- Adds the new option --haploid-contigs
- Improved performance for short-read mode through increased GPU utilization and kernel optimizations.

rna_fq2bam:

• Supports passing --out-chim-type multiple times.

fq2bamfast:

- Improved alignment performance on Hopper GPUs through increased use of <u>DPX</u> instructions.
- Improved performance on multi-GPU runs; for example, on <u>DGX H100</u>.
- Improved error detection for improper FASTQ inputs through --in-fq or --in-se-fq. Previously recorded a utf-8 decode error.
- Additional option to monitor approximate CPU utilization and host memory usage during execution (--monitor-usage).

fq2bam:

- Improved error detection for improper FASTQ inputs through --in-fq or --in-se-fq. Previously recorded a utf-8 decode error.
- Additional option to monitor approximate CPU utilization and host memory usage during execution (--monitor-usage).

minimap2:

• Updated map-pbmm2 preset to match the updated versions of minimap2 (v2.26) and pbmm2 (v1.13.0).

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