

Pipeline Report

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1 Metadata information

1.1 Constants

The following information is not automatically updated in the report:

Build: b38 (hg38)

1.2 Dynamic

The following information is gathered dynamically when a report is generated:

| Library | Sample | Preparation | Gbp |
|-------------|---------|-------------------|--------|
| HALB3002753 | NA12878 | PCR-free-illumina | 181.14 |
| HALB3002754 | HG002 | PCR-free-illumina | 145.87 |
| HALB3002755 | HG003 | PCR-free-illumina | 149.77 |
| HALB3002756 | HG004 | PCR-free-illumina | 138.30 |
| HALB3002757 | HG005 | PCR-free-illumina | 159.85 |
| HALB3003200 | NA12878 | PCR-free-illumina | 167.53 |
| HALB3003201 | HG002 | PCR-free-illumina | 129.81 |
| HALB3003222 | HG003 | PCR-free-illumina | 156.20 |
| HALB3010451 | NA12878 | PacBio-CCS-high | 77.46 |
| HALB3010452 | HG002 | PacBio-CCS-high | 166.22 |
| HALB3010453 | HG003 | PacBio-CCS-high | 129.52 |
| HALB3010454 | HG004 | PacBio-CCS-high | 123.81 |
| HALB3010455 | HG005 | PacBio-CCS-high | 145.34 |
| HALB3010456 | HG006 | PacBio-CCS-high | 122.17 |
| HALB3010457 | HG007 | PacBio-CCS-high | 113.30 |

2 Summarized Small Variant Comparison

This section contains the summarized results of the small variant analysis. For each aligner-caller pair, we ran RTG on the Genome in a Bottle (GIAB) high confidence truth set ($\approx 3.6\text{M}$ variants). The following results are gathered from the RTG ‘summary.txt’ file using the final row (i.e. “None” indicating no variants were filtered) and with the `--all-records` option enabled.

2.1 Preparation: “PCR-free-illumina”

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|------------------|--------------------------|---------------------------|---|----------------|----------------|----------------|
| clinical | sentieon-201808.07 | strelka-2.9.10 | 8 | 0.9923+-0.0014 | 0.9920+-0.0008 | 0.9922+-0.0010 |
| hg38_GIAB_masked | dragmap-1.2.1 | deepvariant-1.2.0 | 8 | 0.9989+-0.0002 | 0.9928+-0.0006 | 0.9959+-0.0004 |
| hg38_GIAB_masked | dragmap-1.2.1 | dnascope-0.5-202010.04 | 8 | 0.9863+-0.0007 | 0.9945+-0.0007 | 0.9904+-0.0007 |
| hg38_GIAB_masked | dragmap-1.2.1 | dnascope-0.5-202010.04-PO | 8 | 0.9984+-0.0002 | 0.9924+-0.0007 | 0.9954+-0.0004 |
| hg38_GIAB_masked | dragmap-1.2.1 | sentieon_gatk4-202010.04 | 8 | 0.9913+-0.0008 | 0.9924+-0.0009 | 0.9918+-0.0008 |
| hg38_GIAB_masked | sentieon-202010.04 | clair3-0.1-r5 | 8 | 0.9973+-0.0003 | 0.9937+-0.0006 | 0.9955+-0.0004 |
| hg38_GIAB_masked | sentieon-202010.04 | deepvariant-1.2.0 | 8 | 0.9988+-0.0003 | 0.9937+-0.0006 | 0.9963+-0.0003 |
| hg38_GIAB_masked | sentieon-202010.04 | dnascope-0.5-202010.04 | 8 | 0.9817+-0.0009 | 0.9947+-0.0006 | 0.9882+-0.0007 |
| hg38_GIAB_masked | sentieon-202010.04 | dnascope-0.5-202010.04-PO | 8 | 0.9980+-0.0002 | 0.9927+-0.0006 | 0.9953+-0.0003 |
| hg38_GIAB_masked | sentieon-202010.04 | octopus-0.7.4 | 8 | 0.9946+-0.0004 | 0.9903+-0.0008 | 0.9924+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04 | sentieon_gatk4-202010.04 | 8 | 0.9865+-0.0009 | 0.9934+-0.0007 | 0.9899+-0.0007 |
| hg38_GIAB_masked | sentieon-202010.04 | strelka-2.9.10 | 8 | 0.9869+-0.0039 | 0.9931+-0.0007 | 0.9900+-0.0022 |
| hg38_GIAB_masked | sentieon-202010.04-recal | clair3-0.1-r5 | 8 | 0.9973+-0.0003 | 0.9927+-0.0007 | 0.9950+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04-recal | deepvariant-1.2.0 | 8 | 0.9987+-0.0002 | 0.9935+-0.0005 | 0.9961+-0.0003 |
| hg38_GIAB_masked | sentieon-202010.04-recal | dnascope-0.5-202010.04 | 8 | 0.9839+-0.0008 | 0.9945+-0.0007 | 0.9892+-0.0007 |
| hg38_GIAB_masked | sentieon-202010.04-recal | dnascope-0.5-202010.04-PO | 8 | 0.9983+-0.0002 | 0.9912+-0.0009 | 0.9948+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04-recal | octopus-0.7.4 | 8 | 0.9947+-0.0004 | 0.9902+-0.0007 | 0.9925+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04-recal | sentieon_gatk4-202010.04 | 8 | 0.9888+-0.0005 | 0.9931+-0.0008 | 0.9909+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04-recal | strelka-2.9.10 | 8 | 0.9905+-0.0016 | 0.9930+-0.0007 | 0.9918+-0.0011 |
| hg38_GIAB_masked | snap-2.0.0 | deepvariant-1.2.0 | 8 | 0.9984+-0.0002 | 0.9937+-0.0005 | 0.9960+-0.0003 |
| hg38_GIAB_masked | snap-2.0.0 | dnascope-0.5-202010.04 | 8 | 0.9820+-0.0006 | 0.9948+-0.0006 | 0.9884+-0.0006 |
| hg38_GIAB_masked | snap-2.0.0 | dnascope-0.5-202010.04-PO | 8 | 0.9973+-0.0002 | 0.9932+-0.0006 | 0.9953+-0.0003 |
| hg38_GIAB_masked | snap-2.0.0 | sentieon_gatk4-202010.04 | 8 | 0.9849+-0.0009 | 0.9939+-0.0007 | 0.9893+-0.0007 |
| hg38_asm5_alt | dragmap-1.2.1 | deepvariant-1.2.0 | 8 | 0.9986+-0.0002 | 0.9709+-0.0008 | 0.9846+-0.0004 |
| hg38_asm5_alt | dragmap-1.2.1 | dnascope-0.5-202010.04 | 8 | 0.9902+-0.0003 | 0.9725+-0.0009 | 0.9813+-0.0006 |

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|---------------|--------------------------|---------------------------|---|----------------|----------------|----------------|
| hg38_asm5_alt | dragmap-1.2.1 | dnascope-0.5-202010.04-PO | 8 | 0.9983+-0.0002 | 0.9710+-0.0011 | 0.9844+-0.0006 |
| hg38_asm5_alt | dragmap-1.2.1 | sentieon_gatk4-202010.04 | 8 | 0.9935+-0.0006 | 0.9702+-0.0011 | 0.9817+-0.0008 |
| hg38_asm5_alt | parabricks-3.5.0 | germline | 1 | 0.9909+-0.0000 | 0.9944+-0.0000 | 0.9927+-0.0000 |
| hg38_asm5_alt | parabricks-3.6.0 | deepvariant | 1 | 0.9986+-0.0000 | 0.9945+-0.0000 | 0.9965+-0.0000 |
| hg38_asm5_alt | sentieon-202010.04 | clair3-0.1-r5 | 8 | 0.9977+-0.0002 | 0.9928+-0.0007 | 0.9952+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04 | deepvariant-1.2.0 | 8 | 0.9988+-0.0002 | 0.9927+-0.0006 | 0.9957+-0.0003 |
| hg38_asm5_alt | sentieon-202010.04 | dnascope-0.5-202010.04 | 8 | 0.9870+-0.0003 | 0.9937+-0.0007 | 0.9903+-0.0005 |
| hg38_asm5_alt | sentieon-202010.04 | dnascope-0.5-202010.04-PO | 8 | 0.9980+-0.0002 | 0.9921+-0.0007 | 0.9950+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04 | octopus-0.7.4 | 8 | 0.9958+-0.0002 | 0.9893+-0.0008 | 0.9926+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04 | sentieon_gatk4-202010.04 | 8 | 0.9899+-0.0005 | 0.9924+-0.0007 | 0.9912+-0.0006 |
| hg38_asm5_alt | sentieon-202010.04 | strelka-2.9.10 | 8 | 0.9890+-0.0038 | 0.9921+-0.0007 | 0.9906+-0.0022 |
| hg38_asm5_alt | sentieon-202010.04-recal | clair3-0.1-r5 | 8 | 0.9977+-0.0002 | 0.9918+-0.0007 | 0.9947+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04-recal | deepvariant-1.2.0 | 8 | 0.9986+-0.0002 | 0.9925+-0.0006 | 0.9956+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04-recal | dnascope-0.5-202010.04 | 8 | 0.9892+-0.0003 | 0.9935+-0.0007 | 0.9913+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04-recal | dnascope-0.5-202010.04-PO | 8 | 0.9983+-0.0002 | 0.9907+-0.0009 | 0.9945+-0.0005 |
| hg38_asm5_alt | sentieon-202010.04-recal | octopus-0.7.4 | 8 | 0.9960+-0.0002 | 0.9893+-0.0008 | 0.9926+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04-recal | sentieon_gatk4-202010.04 | 8 | 0.9922+-0.0002 | 0.9922+-0.0008 | 0.9922+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04-recal | strelka-2.9.10 | 8 | 0.9923+-0.0014 | 0.9920+-0.0008 | 0.9922+-0.0010 |
| hg38_asm5_alt | snap-2.0.0 | deepvariant-1.2.0 | 8 | 0.9984+-0.0002 | 0.9936+-0.0005 | 0.9960+-0.0003 |
| hg38_asm5_alt | snap-2.0.0 | dnascope-0.5-202010.04 | 8 | 0.9843+-0.0005 | 0.9947+-0.0006 | 0.9895+-0.0005 |
| hg38_asm5_alt | snap-2.0.0 | dnascope-0.5-202010.04-PO | 8 | 0.9973+-0.0002 | 0.9932+-0.0006 | 0.9953+-0.0003 |
| hg38_asm5_alt | snap-2.0.0 | sentieon_gatk4-202010.04 | 8 | 0.9867+-0.0008 | 0.9937+-0.0007 | 0.9902+-0.0007 |

2.2 Preparation: “PacBio-CCS-high”

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|------------------|---------------|-----------------------------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | minimap2-2.23 | clair3-0.1-r9 | 7 | 0.9980+-0.0013 | 0.9987+-0.0006 | 0.9983+-0.0010 |
| hg38_GIAB_masked | minimap2-2.23 | deepvariant-1.2.0 | 7 | 0.9976+-0.0018 | 0.9976+-0.0014 | 0.9976+-0.0016 |
| hg38_GIAB_masked | minimap2-2.23 | dnascope_hifi-0.4-202010.04 | 7 | 0.9986+-0.0008 | 0.9986+-0.0006 | 0.9986+-0.0007 |

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|------------------|-------------------------------|-----------------------------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | minimap2-2.23 | peppermargin-dv-r0.7 | 7 | 0.9983+-0.0009 | 0.9984+-0.0006 | 0.9983+-0.0007 |
| hg38_GIAB_masked | pbmm2-1.4.0 | clair3-0.1-r5 | 7 | 0.9984+-0.0013 | 0.9987+-0.0006 | 0.9985+-0.0009 |
| hg38_GIAB_masked | pbmm2-1.4.0 | deepvariant-1.2.0 | 7 | 0.9978+-0.0018 | 0.9975+-0.0014 | 0.9976+-0.0016 |
| hg38_GIAB_masked | pbmm2-1.4.0 | dnascope_hifi-0.4-202010.04 | 7 | 0.9987+-0.0008 | 0.9987+-0.0006 | 0.9987+-0.0007 |
| hg38_GIAB_masked | pbmm2-1.4.0-dvphased-1.2.0 | deepvariant_hp-1.2.0 | 7 | 0.9987+-0.0009 | 0.9984+-0.0006 | 0.9985+-0.0008 |
| hg38_GIAB_masked | pbmm2-1.4.0-peppermargin-r0.4 | deepvariant_hp-1.2.0 | 7 | 0.9987+-0.0008 | 0.9984+-0.0006 | 0.9985+-0.0007 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | clair3-0.1-r5 | 7 | 0.9965+-0.0012 | 0.9983+-0.0006 | 0.9974+-0.0009 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | deepvariant-1.2.0 | 7 | 0.9976+-0.0018 | 0.9976+-0.0014 | 0.9976+-0.0016 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | dnascope_hifi-0.4-202010.04 | 7 | 0.9986+-0.0008 | 0.9986+-0.0006 | 0.9986+-0.0007 |
| hg38_noalt | pbmm2-1.4.0 | dnascope_hifi-0.4-202010.04 | 7 | 0.9987+-0.0008 | 0.9987+-0.0006 | 0.9987+-0.0007 |
| hg38_noalt | sentieon_mm2-202010.04 | dnascope_hifi-0.4-202010.04 | 7 | 0.9986+-0.0008 | 0.9986+-0.0006 | 0.9986+-0.0007 |

3 Summarized SNP/Indel Comparison

This section contains the summarized results of the small variant analysis when stratified by SNP/Indel. Files were filtered using `bcftools filter` to pair down to SNPs or Indels only. For each aligner-caller pair, we ran RTG on the Genome in a Bottle (GIAB) high confidence truth set (≈ 3.6 M variants). The following results are gathered from the RTG ‘summary.txt’ file using the final row (i.e. “None” indicating no variants were filtered) and with the `--all-records` option enabled.

3.1 Preparation: “PCR-free-illumina”

| Reference | Aligner | Caller | Type | N | Precision | Sensitivity | F-measure |
|------------------|--------------------|---------------------------|-------|---|----------------|----------------|----------------|
| clinical | sentieon-201808.07 | strelka-2.9.10 | SNP | 8 | 0.9912+-0.0016 | 0.9922+-0.0007 | 0.9917+-0.0010 |
| | | | Indel | 8 | 0.9885+-0.0009 | 0.9839+-0.0018 | 0.9862+-0.0013 |
| hg38_GIAB_masked | dragmap-1.2.1 | deepvariant-1.2.0 | SNP | 8 | 0.9989+-0.0002 | 0.9927+-0.0007 | 0.9958+-0.0004 |
| | | | Indel | 8 | 0.9964+-0.0004 | 0.9915+-0.0008 | 0.9940+-0.0006 |
| hg38_GIAB_masked | dragmap-1.2.1 | dnascope-0.5-202010.04 | SNP | 8 | 0.9858+-0.0008 | 0.9944+-0.0007 | 0.9900+-0.0007 |
| | | | Indel | 8 | 0.9896+-0.0009 | 0.9945+-0.0007 | 0.9920+-0.0008 |
| hg38_GIAB_masked | dragmap-1.2.1 | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9987+-0.0002 | 0.9923+-0.0006 | 0.9955+-0.0003 |
| | | | Indel | 8 | 0.9966+-0.0004 | 0.9926+-0.0009 | 0.9946+-0.0006 |
| hg38_GIAB_masked | dragmap-1.2.1 | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_GIAB_masked | sentieon-202010.04 | clair3-0.1-r5 | SNP | 8 | 0.9973+-0.0003 | 0.9938+-0.0007 | 0.9955+-0.0004 |
| | | | Indel | 8 | 0.9945+-0.0007 | 0.9910+-0.0010 | 0.9928+-0.0008 |
| hg38_GIAB_masked | sentieon-202010.04 | deepvariant-1.2.0 | SNP | 8 | 0.9988+-0.0003 | 0.9937+-0.0006 | 0.9963+-0.0003 |
| | | | Indel | 8 | 0.9963+-0.0004 | 0.9918+-0.0009 | 0.9940+-0.0006 |
| hg38_GIAB_masked | sentieon-202010.04 | dnascope-0.5-202010.04 | SNP | 8 | 0.9807+-0.0009 | 0.9946+-0.0006 | 0.9876+-0.0007 |
| | | | Indel | 8 | 0.9874+-0.0010 | 0.9944+-0.0007 | 0.9909+-0.0008 |
| hg38_GIAB_masked | sentieon-202010.04 | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9981+-0.0002 | 0.9927+-0.0006 | 0.9954+-0.0003 |
| | | | Indel | 8 | 0.9964+-0.0004 | 0.9926+-0.0008 | 0.9945+-0.0006 |
| hg38_GIAB_masked | sentieon-202010.04 | octopus-0.7.4 | SNP | 8 | 0.9944+-0.0004 | 0.9854+-0.0011 | 0.9899+-0.0006 |
| | | | Indel | 8 | 0.9469+-0.0026 | 0.9612+-0.0023 | 0.9540+-0.0023 |
| hg38_GIAB_masked | sentieon-202010.04 | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_GIAB_masked | sentieon-202010.04 | strelka-2.9.10 | SNP | 8 | 0.9852+-0.0045 | 0.9934+-0.0007 | 0.9893+-0.0025 |
| | | | Indel | 8 | 0.9870+-0.0010 | 0.9844+-0.0018 | 0.9857+-0.0014 |

| Reference | Aligner | Caller | Type | N | Precision | Sensitivity | F-measure |
|------------------|--------------------------|---------------------------|-------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | sentieon-202010.04-recal | clair3-0.1-r5 | SNP | 8 | 0.9974+-0.0003 | 0.9928+-0.0007 | 0.9951+-0.0004 |
| | | | Indel | 8 | 0.9938+-0.0010 | 0.9900+-0.0011 | 0.9919+-0.0010 |
| hg38_GIAB_masked | sentieon-202010.04-recal | deepvariant-1.2.0 | SNP | 8 | 0.9986+-0.0003 | 0.9934+-0.0005 | 0.9960+-0.0003 |
| | | | Indel | 8 | 0.9963+-0.0004 | 0.9921+-0.0008 | 0.9942+-0.0006 |
| hg38_GIAB_masked | sentieon-202010.04-recal | dnascope-0.5-202010.04 | SNP | 8 | 0.9833+-0.0009 | 0.9945+-0.0006 | 0.9888+-0.0007 |
| | | | Indel | 8 | 0.9875+-0.0010 | 0.9937+-0.0010 | 0.9906+-0.0010 |
| hg38_GIAB_masked | sentieon-202010.04-recal | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9986+-0.0002 | 0.9915+-0.0007 | 0.9950+-0.0004 |
| | | | Indel | 8 | 0.9961+-0.0006 | 0.9889+-0.0021 | 0.9925+-0.0014 |
| hg38_GIAB_masked | sentieon-202010.04-recal | octopus-0.7.4 | SNP | 8 | 0.9944+-0.0003 | 0.9854+-0.0011 | 0.9899+-0.0006 |
| | | | Indel | 8 | 0.9476+-0.0025 | 0.9613+-0.0023 | 0.9544+-0.0022 |
| hg38_GIAB_masked | sentieon-202010.04-recal | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_GIAB_masked | sentieon-202010.04-recal | strelka-2.9.10 | SNP | 8 | 0.9893+-0.0018 | 0.9932+-0.0007 | 0.9913+-0.0012 |
| | | | Indel | 8 | 0.9870+-0.0010 | 0.9844+-0.0018 | 0.9857+-0.0014 |
| hg38_GIAB_masked | snap-2.0.0 | deepvariant-1.2.0 | SNP | 8 | 0.9984+-0.0002 | 0.9936+-0.0005 | 0.9960+-0.0003 |
| | | | Indel | 8 | 0.9961+-0.0004 | 0.9921+-0.0009 | 0.9941+-0.0006 |
| hg38_GIAB_masked | snap-2.0.0 | dnascope-0.5-202010.04 | SNP | 8 | 0.9810+-0.0007 | 0.9947+-0.0006 | 0.9878+-0.0006 |
| | | | Indel | 8 | 0.9882+-0.0007 | 0.9946+-0.0007 | 0.9914+-0.0007 |
| hg38_GIAB_masked | snap-2.0.0 | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9974+-0.0002 | 0.9932+-0.0005 | 0.9953+-0.0003 |
| | | | Indel | 8 | 0.9961+-0.0004 | 0.9930+-0.0008 | 0.9946+-0.0006 |
| hg38_GIAB_masked | snap-2.0.0 | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_asm5_alt | dragmap-1.2.1 | deepvariant-1.2.0 | SNP | 8 | 0.9986+-0.0002 | 0.9703+-0.0008 | 0.9842+-0.0004 |
| | | | Indel | 8 | 0.9962+-0.0004 | 0.9732+-0.0011 | 0.9846+-0.0008 |
| hg38_asm5_alt | dragmap-1.2.1 | dnascope-0.5-202010.04 | SNP | 8 | 0.9897+-0.0002 | 0.9719+-0.0010 | 0.9808+-0.0006 |
| | | | Indel | 8 | 0.9932+-0.0008 | 0.9763+-0.0009 | 0.9847+-0.0008 |
| hg38_asm5_alt | dragmap-1.2.1 | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9985+-0.0002 | 0.9704+-0.0011 | 0.9842+-0.0006 |
| | | | Indel | 8 | 0.9967+-0.0005 | 0.9748+-0.0011 | 0.9856+-0.0008 |
| hg38_asm5_alt | dragmap-1.2.1 | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_asm5_alt | parabricks-3.5.0 | germline | SNP | 1 | 0.9905+-0.0000 | 0.9943+-0.0000 | 0.9924+-0.0000 |
| | | | Indel | 1 | 0.9929+-0.0000 | 0.9941+-0.0000 | 0.9935+-0.0000 |
| hg38_asm5_alt | parabricks-3.6.0 | deepvariant | SNP | 1 | 0.9985+-0.0000 | 0.9945+-0.0000 | 0.9965+-0.0000 |

| Reference | Aligner | Caller | Type | N | Precision | Sensitivity | F-measure |
|---------------|--------------------------|---------------------------|-------|---|----------------|----------------|----------------|
| | | | Indel | 1 | 0.9966+-0.0000 | 0.9930+-0.0000 | 0.9948+-0.0000 |
| hg38_asm5_alt | sentieon-202010.04 | clair3-0.1-r5 | SNP | 8 | 0.9976+-0.0002 | 0.9927+-0.0007 | 0.9952+-0.0004 |
| | | | Indel | 8 | 0.9947+-0.0006 | 0.9905+-0.0010 | 0.9926+-0.0008 |
| hg38_asm5_alt | sentieon-202010.04 | deepvariant-1.2.0 | SNP | 8 | 0.9988+-0.0002 | 0.9926+-0.0007 | 0.9957+-0.0003 |
| | | | Indel | 8 | 0.9963+-0.0004 | 0.9913+-0.0009 | 0.9938+-0.0006 |
| hg38_asm5_alt | sentieon-202010.04 | dnascope-0.5-202010.04 | SNP | 8 | 0.9862+-0.0003 | 0.9936+-0.0007 | 0.9898+-0.0005 |
| | | | Indel | 8 | 0.9918+-0.0006 | 0.9939+-0.0007 | 0.9929+-0.0006 |
| hg38_asm5_alt | sentieon-202010.04 | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9982+-0.0002 | 0.9920+-0.0007 | 0.9951+-0.0003 |
| | | | Indel | 8 | 0.9964+-0.0005 | 0.9923+-0.0008 | 0.9944+-0.0006 |
| hg38_asm5_alt | sentieon-202010.04 | octopus-0.7.4 | SNP | 8 | 0.9957+-0.0002 | 0.9844+-0.0012 | 0.9900+-0.0006 |
| | | | Indel | 8 | 0.9478+-0.0025 | 0.9608+-0.0023 | 0.9543+-0.0023 |
| hg38_asm5_alt | sentieon-202010.04 | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_asm5_alt | sentieon-202010.04 | strelka-2.9.10 | SNP | 8 | 0.9874+-0.0043 | 0.9924+-0.0007 | 0.9899+-0.0024 |
| | | | Indel | 8 | 0.9884+-0.0008 | 0.9839+-0.0018 | 0.9861+-0.0013 |
| hg38_asm5_alt | sentieon-202010.04-recal | clair3-0.1-r5 | SNP | 8 | 0.9978+-0.0002 | 0.9918+-0.0007 | 0.9948+-0.0004 |
| | | | Indel | 8 | 0.9941+-0.0009 | 0.9897+-0.0011 | 0.9919+-0.0009 |
| hg38_asm5_alt | sentieon-202010.04-recal | deepvariant-1.2.0 | SNP | 8 | 0.9986+-0.0002 | 0.9924+-0.0006 | 0.9955+-0.0004 |
| | | | Indel | 8 | 0.9963+-0.0004 | 0.9917+-0.0008 | 0.9940+-0.0006 |
| hg38_asm5_alt | sentieon-202010.04-recal | dnascope-0.5-202010.04 | SNP | 8 | 0.9887+-0.0003 | 0.9935+-0.0007 | 0.9911+-0.0004 |
| | | | Indel | 8 | 0.9917+-0.0007 | 0.9931+-0.0010 | 0.9925+-0.0008 |
| hg38_asm5_alt | sentieon-202010.04-recal | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9986+-0.0001 | 0.9909+-0.0008 | 0.9947+-0.0004 |
| | | | Indel | 8 | 0.9961+-0.0007 | 0.9886+-0.0020 | 0.9923+-0.0014 |
| hg38_asm5_alt | sentieon-202010.04-recal | octopus-0.7.4 | SNP | 8 | 0.9958+-0.0002 | 0.9844+-0.0012 | 0.9901+-0.0006 |
| | | | Indel | 8 | 0.9486+-0.0024 | 0.9609+-0.0023 | 0.9547+-0.0022 |
| hg38_asm5_alt | sentieon-202010.04-recal | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| hg38_asm5_alt | sentieon-202010.04-recal | strelka-2.9.10 | SNP | 8 | 0.9912+-0.0016 | 0.9922+-0.0007 | 0.9917+-0.0010 |
| | | | Indel | 8 | 0.9885+-0.0009 | 0.9839+-0.0018 | 0.9862+-0.0013 |
| hg38_asm5_alt | snap-2.0.0 | deepvariant-1.2.0 | SNP | 8 | 0.9984+-0.0002 | 0.9936+-0.0005 | 0.9960+-0.0003 |
| | | | Indel | 8 | 0.9961+-0.0004 | 0.9920+-0.0009 | 0.9940+-0.0006 |
| hg38_asm5_alt | snap-2.0.0 | dnascope-0.5-202010.04 | SNP | 8 | 0.9834+-0.0005 | 0.9946+-0.0006 | 0.9889+-0.0005 |
| | | | Indel | 8 | 0.9899+-0.0007 | 0.9945+-0.0006 | 0.9922+-0.0007 |
| hg38_asm5_alt | snap-2.0.0 | dnascope-0.5-202010.04-PO | SNP | 8 | 0.9974+-0.0002 | 0.9932+-0.0006 | 0.9953+-0.0003 |

| Reference | Aligner | Caller | Type | N | Precision | Sensitivity | F-measure |
|---------------|------------|--------------------------|-------|---|----------------|----------------|----------------|
| | | | Indel | 8 | 0.9960+-0.0004 | 0.9930+-0.0008 | 0.9946+-0.0006 |
| hg38_asm5_alt | snap-2.0.0 | sentieon_gatk4-202010.04 | SNP | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |
| | | | Indel | 8 | nan+-nan | 0.0000+-0.0000 | nan+-nan |

3.2 Preparation: “PacBio-CCS-high”

| Reference | Aligner | Caller | Type | N | Precision | Sensitivity | F-measure |
|------------------|-------------------------------|-----------------------------|-------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | minimap2-2.23 | clair3-0.1-r9 | SNP | 7 | 0.9978+-0.0003 | 0.9994+-0.0001 | 0.9986+-0.0002 |
| | | | Indel | 7 | 0.9880+-0.0089 | 0.9858+-0.0063 | 0.9869+-0.0076 |
| hg38_GIAB_masked | minimap2-2.23 | deepvariant-1.2.0 | SNP | 7 | 0.9984+-0.0003 | 0.9991+-0.0001 | 0.9988+-0.0001 |
| | | | Indel | 7 | 0.9822+-0.0127 | 0.9787+-0.0116 | 0.9805+-0.0121 |
| hg38_GIAB_masked | minimap2-2.23 | dnascope_hifi-0.4-202010.04 | SNP | 7 | 0.9993+-0.0003 | 0.9993+-0.0001 | 0.9993+-0.0001 |
| | | | Indel | 7 | 0.9935+-0.0049 | 0.9932+-0.0050 | 0.9934+-0.0049 |
| hg38_GIAB_masked | minimap2-2.23 | peppermargindv-r0.7 | SNP | 7 | 0.9981+-0.0002 | 0.9991+-0.0002 | 0.9986+-0.0001 |
| | | | Indel | 7 | 0.9896+-0.0057 | 0.9855+-0.0059 | 0.9875+-0.0058 |
| hg38_GIAB_masked | pbmm2-1.4.0 | clair3-0.1-r5 | SNP | 7 | 0.9987+-0.0002 | 0.9993+-0.0002 | 0.9990+-0.0001 |
| | | | Indel | 7 | 0.9903+-0.0087 | 0.9912+-0.0060 | 0.9908+-0.0074 |
| hg38_GIAB_masked | pbmm2-1.4.0 | deepvariant-1.2.0 | SNP | 7 | 0.9990+-0.0002 | 0.9990+-0.0002 | 0.9990+-0.0001 |
| | | | Indel | 7 | 0.9846+-0.0126 | 0.9843+-0.0113 | 0.9844+-0.0119 |
| hg38_GIAB_masked | pbmm2-1.4.0 | dnascope_hifi-0.4-202010.04 | SNP | 7 | 0.9994+-0.0003 | 0.9993+-0.0001 | 0.9993+-0.0001 |
| | | | Indel | 7 | 0.9940+-0.0046 | 0.9940+-0.0045 | 0.9940+-0.0046 |
| hg38_GIAB_masked | pbmm2-1.4.0-dvphased-1.2.0 | deepvariant_hp-1.2.0 | SNP | 7 | 0.9990+-0.0001 | 0.9990+-0.0002 | 0.9990+-0.0001 |
| | | | Indel | 7 | 0.9914+-0.0059 | 0.9910+-0.0057 | 0.9912+-0.0058 |
| hg38_GIAB_masked | pbmm2-1.4.0-peppermargin-r0.4 | deepvariant_hp-1.2.0 | SNP | 7 | 0.9991+-0.0001 | 0.9990+-0.0002 | 0.9990+-0.0001 |
| | | | Indel | 7 | 0.9917+-0.0056 | 0.9912+-0.0056 | 0.9914+-0.0056 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | clair3-0.1-r5 | SNP | 7 | 0.9977+-0.0003 | 0.9987+-0.0002 | 0.9982+-0.0001 |
| | | | Indel | 7 | 0.9773+-0.0081 | 0.9876+-0.0062 | 0.9824+-0.0070 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | deepvariant-1.2.0 | SNP | 7 | 0.9984+-0.0003 | 0.9991+-0.0001 | 0.9988+-0.0001 |
| | | | Indel | 7 | 0.9822+-0.0127 | 0.9787+-0.0116 | 0.9805+-0.0121 |

| Reference | Aligner | Caller | Type | N | Precision | Sensitivity | F-measure |
|------------------|------------------------|-----------------------------|-------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | sentieon_mm2-202010.04 | dnascope_hifi-0.4-202010.04 | SNP | 7 | 0.9993+-0.0003 | 0.9993+-0.0001 | 0.9993+-0.0001 |
| | | | Indel | 7 | 0.9935+-0.0049 | 0.9932+-0.0050 | 0.9934+-0.0049 |
| hg38_noalt | pbmm2-1.4.0 | dnascope_hifi-0.4-202010.04 | SNP | 7 | 0.9994+-0.0003 | 0.9993+-0.0001 | 0.9993+-0.0001 |
| | | | Indel | 7 | 0.9940+-0.0046 | 0.9940+-0.0045 | 0.9940+-0.0046 |
| hg38_noalt | sentieon_mm2-202010.04 | dnascope_hifi-0.4-202010.04 | SNP | 7 | 0.9993+-0.0003 | 0.9993+-0.0001 | 0.9993+-0.0001 |
| | | | Indel | 7 | 0.9935+-0.0049 | 0.9932+-0.0049 | 0.9934+-0.0049 |

4 CMRG Comparison

GIAB recently released a pre-print describing Challenging Medically-Relevant Autosomal Genes (CMRG) benchmark. These regions are harder to capture than the typical GIAB benchmarks. This section contains the summarized results of the small variant analysis, specifically on the CMRG regions. For each aligner-caller pair, we ran RTG on the CMRG high confidence truth set (currently only for HG002 samples). The following results are gathered from the RTG ‘summary.txt’ file using the final row (i.e. “None” indicating no variants were filtered) and with the `--all-records` option enabled.

4.1 Preparation: “PCR-free-illumina”

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|------------------|--------------------------|---------------------------|---|----------------|----------------|----------------|
| clinical | sentieon-201808.07 | strelka-2.9.10 | 2 | 0.9542+-0.0031 | 0.9512+-0.0007 | 0.9527+-0.0019 |
| hg38_GIAB_masked | dragmap-1.2.1 | deepvariant-1.2.0 | 2 | 0.9878+-0.0004 | 0.9601+-0.0005 | 0.9738+-0.0000 |
| hg38_GIAB_masked | dragmap-1.2.1 | dnascope-0.5-202010.04 | 2 | 0.9353+-0.0011 | 0.9659+-0.0008 | 0.9504+-0.0003 |
| hg38_GIAB_masked | dragmap-1.2.1 | dnascope-0.5-202010.04-PO | 2 | 0.9876+-0.0005 | 0.9616+-0.0010 | 0.9744+-0.0007 |
| hg38_GIAB_masked | dragmap-1.2.1 | sentieon_gatk4-202010.04 | 2 | 0.9514+-0.0025 | 0.9527+-0.0008 | 0.9521+-0.0016 |
| hg38_GIAB_masked | sentieon-202010.04 | clair3-0.1-r5 | 2 | 0.9695+-0.0010 | 0.9609+-0.0001 | 0.9651+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04 | deepvariant-1.2.0 | 2 | 0.9870+-0.0002 | 0.9617+-0.0001 | 0.9743+-0.0001 |
| hg38_GIAB_masked | sentieon-202010.04 | dnascope-0.5-202010.04 | 2 | 0.9244+-0.0016 | 0.9657+-0.0009 | 0.9446+-0.0004 |
| hg38_GIAB_masked | sentieon-202010.04 | dnascope-0.5-202010.04-PO | 2 | 0.9848+-0.0002 | 0.9615+-0.0008 | 0.9730+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04 | octopus-0.7.4 | 2 | 0.9735+-0.0017 | 0.9640+-0.0015 | 0.9687+-0.0016 |
| hg38_GIAB_masked | sentieon-202010.04 | sentieon_gatk4-202010.04 | 2 | 0.9406+-0.0014 | 0.9567+-0.0010 | 0.9486+-0.0012 |
| hg38_GIAB_masked | sentieon-202010.04 | strelka-2.9.10 | 2 | 0.9357+-0.0045 | 0.9607+-0.0005 | 0.9481+-0.0025 |
| hg38_GIAB_masked | sentieon-202010.04-recal | clair3-0.1-r5 | 2 | 0.9708+-0.0008 | 0.9578+-0.0000 | 0.9643+-0.0004 |
| hg38_GIAB_masked | sentieon-202010.04-recal | deepvariant-1.2.0 | 2 | 0.9885+-0.0007 | 0.9614+-0.0005 | 0.9748+-0.0005 |
| hg38_GIAB_masked | sentieon-202010.04-recal | dnascope-0.5-202010.04 | 2 | 0.9309+-0.0019 | 0.9648+-0.0007 | 0.9476+-0.0007 |
| hg38_GIAB_masked | sentieon-202010.04-recal | dnascope-0.5-202010.04-PO | 2 | 0.9876+-0.0005 | 0.9569+-0.0008 | 0.9720+-0.0006 |
| hg38_GIAB_masked | sentieon-202010.04-recal | octopus-0.7.4 | 2 | 0.9738+-0.0000 | 0.9634+-0.0007 | 0.9686+-0.0004 |
| hg38_GIAB_masked | sentieon-202010.04-recal | sentieon_gatk4-202010.04 | 2 | 0.9477+-0.0002 | 0.9560+-0.0007 | 0.9518+-0.0002 |
| hg38_GIAB_masked | sentieon-202010.04-recal | strelka-2.9.10 | 2 | 0.9451+-0.0026 | 0.9604+-0.0006 | 0.9526+-0.0017 |
| hg38_GIAB_masked | snap-2.0.0 | deepvariant-1.2.0 | 2 | 0.9869+-0.0009 | 0.9615+-0.0003 | 0.9741+-0.0003 |
| hg38_GIAB_masked | snap-2.0.0 | dnascope-0.5-202010.04 | 2 | 0.9212+-0.0009 | 0.9660+-0.0008 | 0.9430+-0.0001 |
| hg38_GIAB_masked | snap-2.0.0 | dnascope-0.5-202010.04-PO | 2 | 0.9830+-0.0010 | 0.9629+-0.0008 | 0.9729+-0.0009 |
| hg38_GIAB_masked | snap-2.0.0 | sentieon_gatk4-202010.04 | 2 | 0.9343+-0.0011 | 0.9575+-0.0001 | 0.9457+-0.0007 |

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|---------------|--------------------------|---------------------------|---|----------------|----------------|----------------|
| hg38_asm5_alt | dragmap-1.2.1 | deepvariant-1.2.0 | 2 | 0.9854+-0.0003 | 0.8513+-0.0006 | 0.9134+-0.0002 |
| hg38_asm5_alt | dragmap-1.2.1 | dnascope-0.5-202010.04 | 2 | 0.9408+-0.0021 | 0.8565+-0.0008 | 0.8967+-0.0005 |
| hg38_asm5_alt | dragmap-1.2.1 | dnascope-0.5-202010.04-PO | 2 | 0.9827+-0.0004 | 0.8520+-0.0012 | 0.9126+-0.0008 |
| hg38_asm5_alt | dragmap-1.2.1 | sentieon_gatk4-202010.04 | 2 | 0.9587+-0.0018 | 0.8417+-0.0010 | 0.8964+-0.0014 |
| hg38_asm5_alt | sentieon-202010.04 | clair3-0.1-r5 | 2 | 0.9771+-0.0004 | 0.9520+-0.0001 | 0.9644+-0.0003 |
| hg38_asm5_alt | sentieon-202010.04 | deepvariant-1.2.0 | 2 | 0.9882+-0.0006 | 0.9528+-0.0002 | 0.9702+-0.0002 |
| hg38_asm5_alt | sentieon-202010.04 | dnascope-0.5-202010.04 | 2 | 0.9382+-0.0024 | 0.9567+-0.0005 | 0.9474+-0.0010 |
| hg38_asm5_alt | sentieon-202010.04 | dnascope-0.5-202010.04-PO | 2 | 0.9835+-0.0004 | 0.9527+-0.0005 | 0.9679+-0.0004 |
| hg38_asm5_alt | sentieon-202010.04 | octopus-0.7.4 | 2 | 0.9738+-0.0010 | 0.9551+-0.0014 | 0.9643+-0.0012 |
| hg38_asm5_alt | sentieon-202010.04 | sentieon_gatk4-202010.04 | 2 | 0.9512+-0.0013 | 0.9480+-0.0014 | 0.9496+-0.0013 |
| hg38_asm5_alt | sentieon-202010.04 | strelka-2.9.10 | 2 | 0.9449+-0.0050 | 0.9516+-0.0007 | 0.9483+-0.0028 |
| hg38_asm5_alt | sentieon-202010.04-recal | clair3-0.1-r5 | 2 | 0.9785+-0.0005 | 0.9490+-0.0002 | 0.9635+-0.0003 |
| hg38_asm5_alt | sentieon-202010.04-recal | deepvariant-1.2.0 | 2 | 0.9884+-0.0009 | 0.9526+-0.0003 | 0.9702+-0.0006 |
| hg38_asm5_alt | sentieon-202010.04-recal | dnascope-0.5-202010.04 | 2 | 0.9447+-0.0024 | 0.9559+-0.0006 | 0.9503+-0.0008 |
| hg38_asm5_alt | sentieon-202010.04-recal | dnascope-0.5-202010.04-PO | 2 | 0.9859+-0.0007 | 0.9484+-0.0009 | 0.9667+-0.0008 |
| hg38_asm5_alt | sentieon-202010.04-recal | octopus-0.7.4 | 2 | 0.9748+-0.0001 | 0.9550+-0.0013 | 0.9648+-0.0007 |
| hg38_asm5_alt | sentieon-202010.04-recal | sentieon_gatk4-202010.04 | 2 | 0.9587+-0.0002 | 0.9471+-0.0008 | 0.9528+-0.0005 |
| hg38_asm5_alt | sentieon-202010.04-recal | strelka-2.9.10 | 2 | 0.9542+-0.0031 | 0.9512+-0.0007 | 0.9527+-0.0019 |
| hg38_asm5_alt | snap-2.0.0 | deepvariant-1.2.0 | 2 | 0.9869+-0.0007 | 0.9533+-0.0004 | 0.9698+-0.0001 |
| hg38_asm5_alt | snap-2.0.0 | dnascope-0.5-202010.04 | 2 | 0.9318+-0.0014 | 0.9578+-0.0008 | 0.9446+-0.0003 |
| hg38_asm5_alt | snap-2.0.0 | dnascope-0.5-202010.04-PO | 2 | 0.9825+-0.0009 | 0.9546+-0.0008 | 0.9684+-0.0008 |
| hg38_asm5_alt | snap-2.0.0 | sentieon_gatk4-202010.04 | 2 | 0.9431+-0.0007 | 0.9493+-0.0003 | 0.9462+-0.0005 |

4.2 Preparation: “PacBio-CCS-high”

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|------------------|---------------|-----------------------------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | minimap2-2.23 | clair3-0.1-r9 | 1 | 0.9668+-0.0000 | 0.9925+-0.0000 | 0.9795+-0.0000 |
| hg38_GIAB_masked | minimap2-2.23 | deepvariant-1.2.0 | 1 | 0.9792+-0.0000 | 0.9886+-0.0000 | 0.9839+-0.0000 |
| hg38_GIAB_masked | minimap2-2.23 | dnascope_hifi-0.4-202010.04 | 1 | 0.9876+-0.0000 | 0.9894+-0.0000 | 0.9885+-0.0000 |

| Reference | Aligner | Caller | N | Precision | Sensitivity | F-measure |
|------------------|-------------------------------|-----------------------------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | minimap2-2.23 | peppermargindv-r0.7 | 1 | 0.9854+-0.0000 | 0.9908+-0.0000 | 0.9881+-0.0000 |
| hg38_GIAB_masked | pbmm2-1.4.0 | clair3-0.1-r5 | 1 | 0.9687+-0.0000 | 0.9903+-0.0000 | 0.9794+-0.0000 |
| hg38_GIAB_masked | pbmm2-1.4.0 | deepvariant-1.2.0 | 1 | 0.9806+-0.0000 | 0.9874+-0.0000 | 0.9840+-0.0000 |
| hg38_GIAB_masked | pbmm2-1.4.0 | dnascope_hifi-0.4-202010.04 | 1 | 0.9882+-0.0000 | 0.9889+-0.0000 | 0.9886+-0.0000 |
| hg38_GIAB_masked | pbmm2-1.4.0-dvphased-1.2.0 | deepvariant_hp-1.2.0 | 1 | 0.9718+-0.0000 | 0.9892+-0.0000 | 0.9805+-0.0000 |
| hg38_GIAB_masked | pbmm2-1.4.0-peppermargin-r0.4 | deepvariant_hp-1.2.0 | 1 | 0.9741+-0.0000 | 0.9891+-0.0000 | 0.9815+-0.0000 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | clair3-0.1-r5 | 1 | 0.9564+-0.0000 | 0.9919+-0.0000 | 0.9738+-0.0000 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | deepvariant-1.2.0 | 1 | 0.9792+-0.0000 | 0.9886+-0.0000 | 0.9839+-0.0000 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | dnascope_hifi-0.4-202010.04 | 1 | 0.9876+-0.0000 | 0.9894+-0.0000 | 0.9885+-0.0000 |
| hg38_noalt | pbmm2-1.4.0 | dnascope_hifi-0.4-202010.04 | 1 | 0.9864+-0.0000 | 0.9841+-0.0000 | 0.9853+-0.0000 |
| hg38_noalt | sentieon_mm2-202010.04 | dnascope_hifi-0.4-202010.04 | 1 | 0.9857+-0.0000 | 0.9845+-0.0000 | 0.9851+-0.0000 |

5 Summarized Haplotyper Comparison

This section contains the summarized results of the haplotype analysis. For each aligner-haplotyper pair, we attempted to run the haplotyper and gather the resulting prediction. We then calculated the fraction of haplotypers that returned *a* result and the fraction that returned the *correct* result based on our lookup dictionary.

6 Summarized Deletion Comparisons

This section contains the summarized results of the deletion variant analysis. For each aligner-caller pair, we ran Truvari on the Personalis (related to Genome in a Bottle) high confidence deletion truth set ($\approx 2.6\text{K}$ deletions). The following results are gathered from the Truvari “summary.txt” file.

6.1 Preparation: “PCR-free-illumina”

| Reference | Aligner | Caller | N | precision | recall | f1 |
|------------------|--------------------------|-------------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | dragmap-1.2.1 | manta-1.6.0 | 4 | 0.5694+-0.0968 | 0.7414+-0.1496 | 0.6223+-0.0078 |
| hg38_GIAB_masked | sentieon-202010.04 | manta-1.6.0 | 4 | 0.5405+-0.0989 | 0.7585+-0.1441 | 0.6098+-0.0173 |
| hg38_GIAB_masked | sentieon-202010.04-recal | manta-1.6.0 | 4 | 0.5395+-0.0979 | 0.7583+-0.1448 | 0.6092+-0.0166 |
| hg38_GIAB_masked | snap-2.0.0 | manta-1.6.0 | 4 | 0.4962+-0.0946 | 0.7500+-0.1470 | 0.5759+-0.0216 |
| hg38_asm5_alt | dragmap-1.2.1 | manta-1.6.0 | 4 | 0.5851+-0.1027 | 0.7227+-0.1442 | 0.6242+-0.0073 |
| hg38_asm5_alt | sentieon-202010.04 | manta-1.6.0 | 4 | 0.5502+-0.1032 | 0.7555+-0.1427 | 0.6147+-0.0183 |
| hg38_asm5_alt | sentieon-202010.04-recal | manta-1.6.0 | 4 | 0.5492+-0.1020 | 0.7550+-0.1432 | 0.6140+-0.0174 |
| hg38_asm5_alt | snap-2.0.0 | manta-1.6.0 | 4 | 0.4999+-0.0970 | 0.7476+-0.1463 | 0.5773+-0.0224 |

6.2 Preparation: “PacBio-CCS-high”

| Reference | Aligner | Caller | N | precision | recall | f1 |
|------------------|------------------------|------------|---|----------------|----------------|----------------|
| hg38_GIAB_masked | minimap2-2.23 | pbsv-2.6.2 | 2 | 0.3513+-0.1132 | 0.9563+-0.0069 | 0.5029+-0.1210 |
| hg38_GIAB_masked | pbbmm2-1.4.0 | pbsv-2.6.2 | 2 | 0.3720+-0.1221 | 0.9597+-0.0088 | 0.5238+-0.1265 |
| hg38_GIAB_masked | sentieon_mm2-202010.04 | pbsv-2.6.2 | 2 | 0.3513+-0.1137 | 0.9562+-0.0071 | 0.5027+-0.1215 |
| hg38_noalt | pbbmm2-1.4.0 | pbsv-2.6.2 | 2 | 0.3718+-0.1222 | 0.9597+-0.0091 | 0.5235+-0.1266 |
| hg38_noalt | sentieon_mm2-202010.04 | pbsv-2.6.2 | 2 | 0.3515+-0.1139 | 0.9563+-0.0069 | 0.5029+-0.1217 |