Pipeline Report

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Contents

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 (≈3.6M 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_T2T_masked	dragmap-1.2.1	clair3-0.1-r11	8	0.9976+-0.0003	0.9920+-0.0006	0.9948+-0.0003
hg38_T2T_masked	dragmap-1.2.1	clair3-0.1-r9	8	0.9976+-0.0003	0.9925+-0.0006	0.9950+-0.0004
hg38_T2T_masked	dragmap-1.2.1	dnascope-1.0-202112.01-PO	8	0.9984+-0.0003	0.9942+-0.0008	0.9963+-0.0005
hg38_T2T_masked	dragmap-1.2.1	octopus-0.7.4	8	0.9962+-0.0003	0.9899+-0.0008	0.9931+-0.0005
hg38_T2T_masked	dragmap-1.2.1	strelka-2.9.10	8	0.9893+-0.0039	0.9922+-0.0009	0.9907+-0.0023
hg38_T2T_masked	sentieon-202112.01	clair3-0.1-r11	8	0.9974+-0.0003	0.9927+-0.0009	0.9950+-0.0005
hg38_T2T_masked	sentieon-202112.01	clair3-0.1-r9	8	0.9974+-0.0003	0.9936+-0.0006	0.9955+-0.0004
hg38_T2T_masked	sentieon-202112.01	dnascope-1.0-202112.01-PO	8	0.9983+-0.0003	0.9944+-0.0007	0.9963+-0.0005
hg38_T2T_masked	sentieon-202112.01	octopus-0.7.4	8	0.9946+-0.0003	0.9903+-0.0007	0.9924+-0.0005
hg38_T2T_masked	sentieon-202112.01	strelka-2.9.10	8	0.9869+-0.0039	0.9932+-0.0007	0.9900+-0.0022
hg38_T2T_masked	sentieon-202112.01-recal	clair3-0.1-r11	8	0.9974+-0.0003	0.9918+-0.0010	0.9945+-0.0006
hg38_T2T_masked	sentieon-202112.01-recal	clair3-0.1-r9	8	0.9974+-0.0003	0.9924+-0.0010	0.9949+-0.0006
hg38_T2T_masked	sentieon-202112.01-recal	dnascope-1.0-202112.01-PO	8	0.9986+-0.0002	0.9932+-0.0009	0.9959+-0.0005
hg38_T2T_masked	sentieon-202112.01-recal	octopus-0.7.4	8	0.9947+-0.0004	0.9903+-0.0008	0.9925+-0.0005
hg38_T2T_masked	sentieon-202112.01-recal	strelka-2.9.10	8	0.9905+-0.0016	0.9930+-0.0007	0.9918+-0.0011
hg38_T2T_masked	snap-2.0.0	clair3-0.1-r11	8	0.9966+-0.0003	0.9921+-0.0011	0.9943+-0.0007
hg38_T2T_masked	snap-2.0.0	clair3-0.1-r9	8	0.9966+-0.0003	0.9928+-0.0005	0.9947+-0.0004
hg38_T2T_masked	snap-2.0.0	dnascope-1.0-202112.01-PO	8	0.9976+-0.0003	0.9943+-0.0006	0.9959+-0.0004
hg38_T2T_masked	snap-2.0.0	octopus-0.7.4	8	0.9938+-0.0003	0.9907+-0.0007	0.9922+-0.0004
hg38_T2T_masked	snap-2.0.0	strelka-2.9.10	8	0.9861+-0.0042	0.9934+-0.0007	0.9898+-0.0024
hg38_asm5_alt	dragmap-1.2.1	clair3-0.1-r11	8	0.9975+-0.0002	0.9695+-0.0012	0.9833+-0.0007
hg38_asm5_alt	dragmap-1.2.1	clair3-0.1-r9	8	0.9975+-0.0002	0.9705+-0.0008	0.9838+-0.0004
hg38_asm5_alt	dragmap-1.2.1	dnascope-1.0-202112.01-PO	8	0.9978+-0.0002	0.9735+-0.0010	0.9855+-0.0006
hg38_asm5_alt	dragmap-1.2.1	octopus-0.7.4	8	0.9969+-0.0002	0.9681+-0.0011	0.9823+-0.0006

Reference	Aligner	Caller	N	Precision	Sensitivity	F-measure
hg38_asm5_alt	dragmap-1.2.1	strelka-2.9.10	8	0.9902+-0.0038	0.9699+-0.0011	0.9800+-0.0023
hg38_asm5_alt	sentieon-202112.01	clair3-0.1-r11	8	0.9977+-0.0002	0.9922+-0.0009	0.9949+-0.0005
hg38_asm5_alt	sentieon-202112.01	clair3-0.1-r9	8	0.9977+-0.0002	0.9926+-0.0007	0.9951+-0.0004
hg38_asm5_alt	sentieon-202112.01	dnascope-1.0-202112.01-PO	8	0.9983+-0.0002	0.9938+-0.0007	0.9961+-0.0004
hg38_asm5_alt	sentieon-202112.01	octopus-0.7.4	8	0.9958+-0.0002	0.9893+-0.0008	0.9926+-0.0004
hg38_asm5_alt	sentieon-202112.01	strelka-2.9.10	8	0.9890+-0.0038	0.9921+-0.0007	0.9906+-0.0022
hg38_asm5_alt	sentieon-202112.01-recal	clair3-0.1-r11	8	0.9978+-0.0002	0.9911+-0.0008	0.9944+-0.0005
hg38_asm5_alt	sentieon-202112.01-recal	clair3-0.1-r9	8	0.9978+-0.0002	0.9916+-0.0008	0.9947+-0.0004
hg38_asm5_alt	sentieon-202112.01-recal	dnascope-1.0-202112.01-PO	8	0.9986+-0.0002	0.9926+-0.0008	0.9956+-0.0005
hg38_asm5_alt	sentieon-202112.01-recal	octopus-0.7.4	8	0.9960+-0.0002	0.9893+-0.0008	0.9926+-0.0004
hg38_asm5_alt	sentieon-202112.01-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	clair3-0.1-r11	8	0.9967+-0.0003	0.9920+-0.0008	0.9944+-0.0005
hg38_asm5_alt	snap-2.0.0	clair3-0.1-r9	8	0.9967+-0.0003	0.9927+-0.0005	0.9947+-0.0004
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-1.0-202112.01-PO	8	0.9976+-0.0003	0.9941+-0.0006	0.9959+-0.0004
hg38_asm5_alt	snap-2.0.0	octopus-0.7.4	8	0.9944+-0.0003	0.9905+-0.0007	0.9925+-0.0004
hg38_asm5_alt	snap-2.0.0	strelka-2.9.10	8	0.9868+-0.0042	0.9932+-0.0007	0.9900+-0.0024

2.2 Preparation: "PacBio-CCS-high"

Reference	Aligner	Caller	N	Precision	Sensitivity	F-measure
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r11	7	0.9980+-0.0013	0.9987+-0.0006	0.9984+-0.0010
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r9	7	0.9980+-0.0013	0.9987+-0.0006	0.9983+-0.0010
hg38_T2T_masked	minimap2-2.23	dnascope_hifi-0.4-202112.01	7	0.9986+-0.0008	0.9986+-0.0006	0.9986+-0.0007
hg38_T2T_masked	minimap2-2.23	peppermargindv-r0.8	7	0.9983+-0.0009	0.9985+-0.0005	0.9984+-0.0007
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r11	7	0.9984+-0.0013	0.9987+-0.0006	0.9985+-0.0009
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r9	7	0.9984+-0.0013	0.9987+-0.0006	0.9985+-0.0009
hg38_T2T_masked	pbmm2-1.7.0	dnascope_hifi-0.4-202112.01	7	0.9987+-0.0008	0.9987+-0.0005	0.9987+-0.0007
hg38_T2T_masked	pbmm2-1.7.0	peppermargindv-r0.8	7	0.9985+-0.0009	0.9984+-0.0005	0.9985+-0.0007

Reference	Aligner	Caller	N	Precision	Sensitivity	F-measure
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r11	7	0.9980+-0.0013	0.9987+-0.0006	0.9983+-0.0010
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r9	7	0.9980+-0.0013	0.9987+-0.0006	0.9983+-0.0010
hg38_T2T_masked	sentieon_mm2-202112.01	dnascope_hifi-0.4-202112.01	7	0.9986+-0.0008	0.9986+-0.0006	0.9986+-0.0007
hg38_T2T_masked	sentieon_mm2-202112.01	peppermargindv-r0.8	7	0.9983+-0.0009	0.9985+-0.0005	0.9984+-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 ($\approx$3.6M 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_T2T_masked	dragmap-1.2.1	clair3-0.1-r11	SNP	8	0.9976+-0.0003	0.9920+-0.0006	0.9948+-0.0003
			Indel	8	0.9949+-0.0006	0.9890+-0.0010	0.9920+-0.0008
hg38_T2T_masked	dragmap-1.2.1	clair3-0.1-r9	SNP	8	0.9976+-0.0003	0.9925+-0.0006	0.9950+-0.0004
			Indel	8	0.9949+-0.0006	0.9896+-0.0011	0.9922+-0.0008
hg38_T2T_masked	dragmap-1.2.1	dnascope-1.0-202112.01-PO	SNP	8	0.9986+-0.0002	0.9940+-0.0008	0.9963+-0.0005
			Indel	8	0.9968+-0.0005	0.9946+-0.0008	0.9957+-0.0006
hg38_T2T_masked	dragmap-1.2.1	octopus-0.7.4	SNP	8	0.9961+-0.0003	0.9851+-0.0012	0.9906+-0.0006
			Indel	8	0.9476+-0.0026	0.9612+-0.0024	0.9544+-0.0024
hg38_T2T_masked	dragmap-1.2.1	strelka-2.9.10	SNP	8	0.9878+-0.0045	0.9924+-0.0009	0.9900+-0.0025
			Indel	8	0.9883+-0.0010	0.9841+-0.0019	0.9862+-0.0014
hg38_T2T_masked	sentieon-202112.01	clair3-0.1-r11	SNP	8	0.9973+-0.0003	0.9929+-0.0009	0.9951+-0.0005
			Indel	8	0.9948+-0.0006	0.9889+-0.0015	0.9918+-0.0010
hg38_T2T_masked	sentieon-202112.01	clair3-0.1-r9	SNP	8	0.9973+-0.0003	0.9938+-0.0006	0.9955+-0.0004
			Indel	8	0.9948+-0.0006	0.9900+-0.0012	0.9924+-0.0009
hg38_T2T_masked	sentieon-202112.01	dnascope-1.0-202112.01-PO	SNP	8	0.9984+-0.0002	0.9942+-0.0007	0.9963+-0.0004
			Indel	8	0.9968+-0.0005	0.9946+-0.0008	0.9957+-0.0006
hg38_T2T_masked	sentieon-202112.01	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_T2T_masked	sentieon-202112.01	strelka-2.9.10	SNP	8	0.9852+-0.0045	0.9935+-0.0006	0.9893+-0.0025
			Indel	8	0.9870+-0.0010	0.9844+-0.0018	0.9857+-0.0014
hg38_T2T_masked	sentieon-202112.01-recal	clair3-0.1-r11	SNP	8	0.9974+-0.0003	0.9919+-0.0009	0.9947+-0.0005
			Indel	8	0.9942+-0.0008	0.9881+-0.0016	0.9911+-0.0011

Reference	Aligner	Caller	Type	N	Precision	Sensitivity	F-measure
hg38_T2T_masked	sentieon-202112.01-recal	clair3-0.1-r9	SNP	8	0.9974+-0.0003	0.9927+-0.0009	0.9950+-0.0005
			Indel	8	0.9942+-0.0008	0.9889+-0.0016	0.9915+-0.0012
hg38_T2T_masked	sentieon-202112.01-recal	dnascope-1.0-202112.01-PO	SNP	8	0.9987+-0.0002	0.9931+-0.0009	0.9959+-0.0005
			Indel	8	0.9967+-0.0006	0.9927+-0.0014	0.9947+-0.0010
hg38_T2T_masked	sentieon-202112.01-recal	octopus-0.7.4	SNP	8	0.9944+-0.0004	0.9854+-0.0011	0.9899+-0.0006
			Indel	8	0.9476+-0.0025	0.9613+-0.0023	0.9544+-0.0023
hg38_T2T_masked	sentieon-202112.01-recal	strelka-2.9.10	SNP	8	0.9893+-0.0018	0.9933+-0.0006	0.9913+-0.0012
			Indel	8	0.9870+-0.0010	0.9844+-0.0018	0.9857+-0.0014
hg38_T2T_masked	snap-2.0.0	clair3-0.1-r11	SNP	8	0.9964+-0.0003	0.9921+-0.0011	0.9942+-0.0006
			Indel	8	0.9943+-0.0006	0.9895+-0.0018	0.9919+-0.0012
hg38_T2T_masked	snap-2.0.0	clair3-0.1-r9	SNP	8	0.9964+-0.0003	0.9929+-0.0005	0.9946+-0.0003
			Indel	8	0.9943+-0.0006	0.9903+-0.0012	0.9923+-0.0009
hg38_T2T_masked	snap-2.0.0	dnascope-1.0-202112.01-PO	SNP	8	0.9977+-0.0002	0.9941+-0.0006	0.9959+-0.0004
			Indel	8	0.9962+-0.0004	0.9945+-0.0007	0.9953+-0.0005
hg38_T2T_masked	snap-2.0.0	octopus-0.7.4	SNP	8	0.9936+-0.0003	0.9858+-0.0011	0.9897+-0.0006
			Indel	8	0.9463+-0.0025	0.9609+-0.0023	0.9535+-0.0023
hg38_T2T_masked	snap-2.0.0	strelka-2.9.10	SNP	8	0.9845+-0.0048	0.9937+-0.0006	0.9891+-0.0026
			Indel	8	0.9853+-0.0009	0.9849+-0.0018	0.9851+-0.0014
hg38_asm5_alt	dragmap-1.2.1	clair3-0.1-r11	SNP	8	0.9975+-0.0002	0.9691+-0.0012	0.9831+-0.0006
			Indel	8	0.9948+-0.0006	0.9703+-0.0019	0.9824+-0.0012
hg38_asm5_alt	dragmap-1.2.1	clair3-0.1-r9	SNP	8	0.9975+-0.0002	0.9700+-0.0008	0.9836+-0.0004
			Indel	8	0.9949+-0.0006	0.9714+-0.0014	0.9830+-0.0010
hg38_asm5_alt	dragmap-1.2.1	dnascope-1.0-202112.01-PO	SNP	8	0.9980+-0.0002	0.9728+-0.0010	0.9853+-0.0006
			Indel	8	0.9966+-0.0006	0.9776+-0.0010	0.9870+-0.0007
hg38_asm5_alt	dragmap-1.2.1	octopus-0.7.4	SNP	8	0.9968+-0.0002	0.9628+-0.0016	0.9796+-0.0008
			Indel	8	0.9482+-0.0027	0.9436+-0.0024	0.9459+-0.0024
hg38_asm5_alt	dragmap-1.2.1	strelka-2.9.10	SNP	8	0.9886+-0.0044	0.9696+-0.0011	0.9790+-0.0026
			Indel	8	0.9894+-0.0009	0.9656+-0.0021	0.9773+-0.0015
hg38_asm5_alt	sentieon-202112.01	clair3-0.1-r11	SNP	8	0.9976+-0.0002	0.9923+-0.0009	0.9950+-0.0005
			Indel	8	0.9950+-0.0005	0.9890+-0.0012	0.9920+-0.0008
hg38_asm5_alt	sentieon-202112.01	clair3-0.1-r9	SNP	8	0.9976+-0.0002	0.9927+-0.0007	0.9952+-0.0004
			Indel	8	0.9950+-0.0005	0.9894+-0.0012	0.9922+-0.0009
hg38_asm5_alt	sentieon-202112.01	dnascope-1.0-202112.01-PO	SNP	8	0.9984+-0.0002	0.9936+-0.0007	0.9960+-0.0004

Reference	Aligner	Caller	Type	N	Precision	Sensitivity	F-measure
			Indel	8	0.9970+-0.0004	0.9943+-0.0007	0.9957+-0.0006
hg38_asm5_alt	sentieon-202112.01	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-202112.01	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-202112.01-recal	clair3-0.1-r11	SNP	8	0.9978+-0.0002	0.9912+-0.0008	0.9945+-0.0004
			Indel	8	0.9945+-0.0008	0.9880+-0.0011	0.9912+-0.0009
hg38_asm5_alt	sentieon-202112.01-recal	clair3-0.1-r9	SNP	8	0.9978+-0.0002	0.9917+-0.0008	0.9948+-0.0004
			Indel	8	0.9945+-0.0008	0.9886+-0.0013	0.9916+-0.0010
hg38_asm5_alt	sentieon-202112.01-recal	dnascope-1.0-202112.01-PO	SNP	8	0.9988+-0.0001	0.9925+-0.0008	0.9956+-0.0004
			Indel	8	0.9968+-0.0006	0.9923+-0.0014	0.9946+-0.0010
hg38_asm5_alt	sentieon-202112.01-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-202112.01-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	clair3-0.1-r11	SNP	8	0.9966+-0.0003	0.9920+-0.0008	0.9943+-0.0005
			Indel	8	0.9944+-0.0006	0.9895+-0.0015	0.9919+-0.0010
hg38_asm5_alt	snap-2.0.0	clair3-0.1-r9	SNP	8	0.9966+-0.0003	0.9927+-0.0005	0.9946+-0.0003
			Indel	8	0.9944+-0.0006	0.9902+-0.0011	0.9923+-0.0008
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-1.0-202112.01-PO	SNP	8	0.9977+-0.0002	0.9939+-0.0006	0.9958+-0.0004
			Indel	8	0.9963+-0.0004	0.9944+-0.0007	0.9953+-0.0005
hg38_asm5_alt	snap-2.0.0	octopus-0.7.4	SNP	8	0.9942+-0.0003	0.9856+-0.0011	0.9899+-0.0006
			Indel	8	0.9467+-0.0026	0.9608+-0.0023	0.9537+-0.0023
hg38_asm5_alt	snap-2.0.0	strelka-2.9.10	SNP	8	0.9852+-0.0048	0.9935+-0.0006	0.9894+-0.0026
			Indel	8	0.9860+-0.0009	0.9848+-0.0018	0.9854+-0.0013

3.2 Preparation: "PacBio-CCS-high"

Reference	Aligner	Caller	Type	N	Precision	Sensitivity	F-measure
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r11	SNP	7	0.9979+-0.0003	0.9994+-0.0001	0.9986+-0.0001
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r11	Indel	7	0.9880+-0.0089	0.9859+-0.0064	0.9869+-0.0076
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r9	SNP	7	0.9978+-0.0003	0.9994+-0.0001	0.9986+-0.0001
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r9	Indel	7	0.9880+-0.0088	0.9859+-0.0063	0.9870+-0.0075
hg38_T2T_masked	minimap2-2.23	dnascope_hifi-0.4-202112.01	SNP	7	0.9993+-0.0002	0.9993+-0.0001	0.9993+-0.0001
hg38_T2T_masked	minimap2-2.23	dnascope_hifi-0.4-202112.01	Indel	7	0.9935+-0.0049	0.9932+-0.0050	0.9934+-0.0049
hg38_T2T_masked	minimap2-2.23	peppermargindv-r0.8	SNP	7	0.9983+-0.0003	0.9990+-0.0001	0.9986+-0.0001
hg38_T2T_masked	minimap2-2.23	peppermargindv-r0.8	Indel	7	0.9886+-0.0056	0.9858+-0.0055	0.9872+-0.0055
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r11	SNP	7	0.9987+-0.0002	0.9993+-0.0002	0.9990+-0.0001
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r11	Indel	7	0.9903+-0.0087	0.9913+-0.0059	0.9908+-0.0073
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r9	SNP	7	0.9987+-0.0002	0.9993+-0.0002	0.9990+-0.0001
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r9	Indel	7	0.9903+-0.0087	0.9913+-0.0059	0.9908+-0.0073
hg38_T2T_masked	pbmm2-1.7.0	dnascope_hifi-0.4-202112.01	SNP	7	0.9993+-0.0003	0.9993+-0.0001	0.9994+-0.0001
hg38_T2T_masked	pbmm2-1.7.0	dnascope_hifi-0.4-202112.01	Indel	7	0.9940+-0.0046	0.9940+-0.0045	0.9940+-0.0046
hg38_T2T_masked	pbmm2-1.7.0	peppermargindv-r0.8	SNP	7	0.9989+-0.0002	0.9989+-0.0002	0.9989+-0.0001
hg38_T2T_masked	pbmm2-1.7.0	peppermargindv-r0.8	Indel	7	0.9908+-0.0055	0.9914+-0.0049	0.9911+-0.0052
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r11	SNP	7	0.9979+-0.0003	0.9994+-0.0001	0.9986+-0.0001
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r11	Indel	7	0.9880+-0.0089	0.9859+-0.0064	0.9869+-0.0076
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r9	SNP	7	0.9979+-0.0003	0.9994+-0.0001	0.9986+-0.0001
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r9	Indel	7	0.9880+-0.0089	0.9858+-0.0063	0.9869+-0.0076
hg38_T2T_masked	sentieon_mm2-202112.01	dnascope_hifi-0.4-202112.01	SNP	7	0.9993+-0.0002	0.9993+-0.0001	0.9993+-0.0001
hg38_T2T_masked	sentieon_mm2-202112.01	dnascope_hifi-0.4-202112.01	Indel	7	0.9935+-0.0049	0.9932+-0.0050	0.9934+-0.0049
hg38_T2T_masked	sentieon_mm2-202112.01	peppermargindv-r0.8	SNP	7	0.9983+-0.0003	0.9990+-0.0001	0.9986+-0.0001
hg38_T2T_masked	sentieon_mm2-202112.01	peppermargindv-r0.8	Indel	7	0.9886+-0.0056	0.9858+-0.0055	0.9872+-0.0055

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_T2T_masked	dragmap-1.2.1	clair3-0.1-r11	2	0.9708+-0.0001	0.9592+-0.0002	0.9650+-0.0000
hg38_T2T_masked	dragmap-1.2.1	clair3-0.1-r9	2	0.9708+-0.0001	0.9597+-0.0002	0.9652+-0.0002
hg38_T2T_masked	dragmap-1.2.1	dnascope-1.0-202112.01-PO	2	0.9753+-0.0001	0.9668+-0.0007	0.9710+-0.0004
hg38_T2T_masked	dragmap-1.2.1	octopus-0.7.4	2	0.9804+-0.0007	0.9634+-0.0009	0.9718+-0.0008
hg38_T2T_masked	dragmap-1.2.1	strelka-2.9.10	2	0.9405+-0.0033	0.9562+-0.0008	0.9482+-0.0021
hg38_T2T_masked	sentieon-202112.01	clair3-0.1-r11	2	0.9696+-0.0004	0.9594+-0.0010	0.9645+-0.0004
hg38_T2T_masked	sentieon-202112.01	clair3-0.1-r9	2	0.9697+-0.0004	0.9605+-0.0002	0.9650+-0.0000
hg38_T2T_masked	sentieon-202112.01	dnascope-1.0-202112.01-PO	2	0.9750+-0.0009	0.9661+-0.0015	0.9706+-0.0011
hg38_T2T_masked	sentieon-202112.01	octopus-0.7.4	2	0.9734+-0.0016	0.9640+-0.0015	0.9687+-0.0016
hg38_T2T_masked	sentieon-202112.01	strelka-2.9.10	2	0.9357+-0.0045	0.9608+-0.0006	0.9481+-0.0025
hg38_T2T_masked	sentieon-202112.01-recal	clair3-0.1-r11	2	0.9707+-0.0005	0.9564+-0.0010	0.9634+-0.0003
hg38_T2T_masked	sentieon-202112.01-recal	clair3-0.1-r9	2	0.9707+-0.0005	0.9575+-0.0001	0.9640+-0.0001
hg38_T2T_masked	sentieon-202112.01-recal	dnascope-1.0-202112.01-PO	2	0.9767+-0.0015	0.9626+-0.0015	0.9695+-0.0015
hg38_T2T_masked	sentieon-202112.01-recal	octopus-0.7.4	2	0.9738+-0.0001	0.9635+-0.0007	0.9687+-0.0005
hg38_T2T_masked	sentieon-202112.01-recal	strelka-2.9.10	2	0.9449+-0.0026	0.9604+-0.0006	0.9526+-0.0017
hg38_T2T_masked	snap-2.0.0	clair3-0.1-r11	2	0.9644+-0.0016	0.9586+-0.0011	0.9614+-0.0014
hg38_T2T_masked	snap-2.0.0	clair3-0.1-r9	2	0.9645+-0.0015	0.9601+-0.0002	0.9623+-0.0009
hg38_T2T_masked	snap-2.0.0	dnascope-1.0-202112.01-PO	2	0.9704+-0.0012	0.9654+-0.0006	0.9679+-0.0009
hg38_T2T_masked	snap-2.0.0	octopus-0.7.4	2	0.9670+-0.0018	0.9652+-0.0007	0.9661+-0.0005
hg38_T2T_masked	snap-2.0.0	strelka-2.9.10	2	0.9269+-0.0055	0.9606+-0.0006	0.9435+-0.0031
hg38_asm5_alt	dragmap-1.2.1	clair3-0.1-r11	2	0.9750+-0.0001	0.8495+-0.0001	0.9080+-0.0001
hg38_asm5_alt	dragmap-1.2.1	clair3-0.1-r9	2	0.9750+-0.0001	0.8511+-0.0002	0.9088+-0.0002

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-1.0-202112.01-PO	2	0.9780+-0.0001	0.8656+-0.0008	0.9183+-0.0004
hg38_asm5_alt	dragmap-1.2.1	octopus-0.7.4	2	0.9747+-0.0000	0.8539+-0.0012	0.9103+-0.0007
hg38_asm5_alt	dragmap-1.2.1	strelka-2.9.10	2	0.9430+-0.0029	0.8447+-0.0010	0.8911+-0.0019
hg38_asm5_alt	sentieon-202112.01	clair3-0.1-r11	2	0.9777+-0.0004	0.9517+-0.0000	0.9646+-0.0002
hg38_asm5_alt	sentieon-202112.01	clair3-0.1-r9	2	0.9777+-0.0004	0.9517+-0.0000	0.9646+-0.0002
hg38_asm5_alt	sentieon-202112.01	dnascope-1.0-202112.01-PO	2	0.9831+-0.0005	0.9622+-0.0012	0.9726+-0.0009
hg38_asm5_alt	sentieon-202112.01	octopus-0.7.4	2	0.9738+-0.0010	0.9551+-0.0014	0.9644+-0.0013
hg38_asm5_alt	sentieon-202112.01	strelka-2.9.10	2	0.9449+-0.0050	0.9516+-0.0007	0.9483+-0.0028
hg38_asm5_alt	sentieon-202112.01-recal	clair3-0.1-r11	2	0.9789+-0.0002	0.9486+-0.0001	0.9635+-0.0002
hg38_asm5_alt	sentieon-202112.01-recal	clair3-0.1-r9	2	0.9789+-0.0002	0.9486+-0.0001	0.9635+-0.0002
hg38_asm5_alt	sentieon-202112.01-recal	dnascope-1.0-202112.01-PO	2	0.9847+-0.0012	0.9589+-0.0010	0.9717+-0.0011
hg38_asm5_alt	sentieon-202112.01-recal	octopus-0.7.4	2	0.9748+-0.0000	0.9550+-0.0013	0.9648+-0.0007
hg38_asm5_alt	sentieon-202112.01-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	clair3-0.1-r11	2	0.9724+-0.0010	0.9505+-0.0005	0.9612+-0.0007
hg38_asm5_alt	snap-2.0.0	clair3-0.1-r9	2	0.9724+-0.0010	0.9520+-0.0002	0.9621+-0.0006
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-1.0-202112.01-PO	2	0.9802+-0.0010	0.9583+-0.0003	0.9691+-0.0006
hg38_asm5_alt	snap-2.0.0	octopus-0.7.4	2	0.9666+-0.0018	0.9575+-0.0011	0.9620+-0.0003
hg38_asm5_alt	snap-2.0.0	strelka-2.9.10	2	0.9335+-0.0053	0.9525+-0.0008	0.9429+-0.0031

4.2 Preparation: "PacBio-CCS-high"

Reference	Aligner	Caller	N	Precision	Sensitivity	F-measure
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r11	1	0.9661+-0.0000	0.9927+-0.0000	0.9792+-0.0000
hg38_T2T_masked	minimap2-2.23	clair3-0.1-r9	1	0.9671+-0.0000	0.9927+-0.0000	0.9797+-0.0000
hg38_T2T_masked	minimap2-2.23	dnascope_hifi-0.4-202112.01	1	0.9876+-0.0000	0.9894+-0.0000	0.9885+-0.0000
hg38_T2T_masked	minimap2-2.23	peppermargindv-r0.8	1	0.9862+-0.0000	0.9908+-0.0000	0.9885+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r11	1	0.9681+-0.0000	0.9902+-0.0000	0.9790+-0.0000

Reference	Aligner	Caller	N	Precision	Sensitivity	F-measure
hg38_T2T_masked	pbmm2-1.7.0	clair3-0.1-r9	1	0.9689+-0.0000	0.9904+-0.0000	0.9795+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	dnascope_hifi-0.4-202112.01	1	0.9881+-0.0000	0.9889+-0.0000	0.9885+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	peppermargindv-r0.8	1	0.9806+-0.0000	0.9888+-0.0000	0.9847+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r11	1	0.9661+-0.0000	0.9928+-0.0000	0.9792+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	clair3-0.1-r9	1	0.9669+-0.0000	0.9927+-0.0000	0.9796+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	dnascope_hifi-0.4-202112.01	1	0.9876+-0.0000	0.9894+-0.0000	0.9885+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	peppermargindv-r0.8	1	0.9860+-0.0000	0.9908+-0.0000	0.9884+-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. "Output" is the percentage that generated an output file. This is most commonly below 100% when a tool crashes while trying to run. "Computed" is the percentage that generated a prediction in the output file. This is most commonly below 100% when a tool successfully runs, but it does not generate a prediction in the output file (e.g. "None"). "Matching" is the percentage that generated the correct haplotype prediction based on our lookup table. These haplotyping tools often assume a particular data type, and are not designed to work under all of the conditions tested by this benchmark.

5.1 Preparation: "PCR-free-illumina"

Reference	Aligner	Haplotyper	N	Output (%)	Computed (%)	Matching (%)
hg38_T2T_masked	dragmap-1.2.1	cyrius-1.1.1	8	100.0	75.0	75.0
hg38_T2T_masked	sentieon-202112.01	cyrius-1.1.1	8	100.0	87.5	87.5
hg38_T2T_masked	sentieon-202112.01-recal	cyrius-1.1.1	8	100.0	87.5	87.5
hg38_T2T_masked	snap-2.0.0	cyrius-1.1.1	8	100.0	87.5	87.5
hg38_asm5_alt	dragmap-1.2.1	cyrius-1.1.1	8	25.0	0.0	0.0
hg38_asm5_alt	sentieon-202112.01	cyrius-1.1.1	8	100.0	100.0	100.0
hg38_asm5_alt	sentieon-202112.01-recal	cyrius-1.1.1	8	100.0	100.0	100.0
hg38_asm5_alt	snap-2.0.0	cyrius-1.1.1	8	100.0	87.5	87.5

5.2 Preparation: "PacBio-CCS-high"

Reference	Aligner	Haplotyper	N	Output (%)	Computed (%)	Matching (%)
hg38_T2T_masked	minimap2-2.23	cyrius-1.1.1	5	100.0	20.0	20.0
hg38_T2T_masked	pbmm2-1.7.0	cyrius-1.1.1	5	100.0	40.0	40.0

Reference	Aligner	Haplotyper	N	Output (%)	Computed (%)	Matching (%)
hg38_T2T_masked	sentieon_mm2-202112.01	cyrius-1.1.1	5	100.0	20.0	20.0

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 corresponding truth set with these options: "`--pctsim 0.0 --sizemax 1000000000`". The following results are gathered from the Truvari "summary.txt" file.

6.1 Restricted calls

These evaluations are **RESTRICTED**, meaning that a BED file was used to limit the analysis regions, typically to "high-confidence" regions associated with the benchmark. This corresponds to the "`--includebed`" option in Truvari. As a result, benchmark sets that were not released with a corresponding BED file are excluded from this analysis. At this time, only the HG002 GIAB v0.6 benchmark is included in this analysis.

6.1.1 Preparation: "PCR-free-illumina"

Reference	Aligner	Caller	N	precision	recall	f1
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.10	2	0.4213+-0.2593	0.7275+-0.0092	0.4823+-0.2167
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.10-PO	2	0.9016+-0.0233	0.7096+-0.0038	0.7940+-0.0066
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.4	2	0.4324+-0.2752	0.7246+-0.0085	0.4853+-0.2265
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.4-PO	2	0.9105+-0.0212	0.7073+-0.0035	0.7960+-0.0059
hg38_T2T_masked	dragmap-1.2.1	manta-1.6.0	2	0.9561+-0.0011	0.6677+-0.0188	0.7862+-0.0127
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.10	2	0.4675+-0.2723	0.6866+-0.0055	0.5069+-0.2023
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.10-PO	2	0.9019+-0.0201	0.6656+-0.0023	0.7658+-0.0057
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.4	2	0.4682+-0.2839	0.6704+-0.0047	0.4979+-0.2083
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.4-PO	2	0.9025+-0.0211	0.6508+-0.0014	0.7562+-0.0064
hg38_T2T_masked	sentieon-202112.01	manta-1.6.0	2	0.9529+-0.0007	0.6909+-0.0153	0.8009+-0.0101
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.10	2	0.4723+-0.2752	0.6855+-0.0054	0.5095+-0.2028
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.10-PO	2	0.8991+-0.0211	0.6657+-0.0024	0.7649+-0.0060
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.4	2	0.4737+-0.2870	0.6689+-0.0085	0.4999+-0.2072
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.4-PO	2	0.9031+-0.0172	0.6516+-0.0060	0.7568+-0.0020
hg38_T2T_masked	sentieon-202112.01-recal	manta-1.6.0	2	0.9518+-0.0009	0.6909+-0.0149	0.8005+-0.0097
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.10	2	0.6886+-0.0520	0.6817+-0.0126	0.6836+-0.0194
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.10-PO	2	0.9401+-0.0050	0.6418+-0.0104	0.7628+-0.0057
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.4	2	0.7011+-0.0512	0.6637+-0.0083	0.6806+-0.0199
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.4-PO	2	0.9420+-0.0040	0.6266+-0.0043	0.7526+-0.0018
hg38_T2T_masked	snap-2.0.0	manta-1.6.0	2	0.9490+-0.0008	0.6805+-0.0128	0.7925+-0.0084
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.10	2	0.4267+-0.2640	0.7112+-0.0083	0.4810+-0.2157

Reference	Aligner	Caller	N	precision	recall	f1
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.10-PO	2	0.9052+-0.0229	0.6944+-0.0032	0.7857+-0.0066
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.4	2	0.4355+-0.2775	0.7088+-0.0097	0.4825+-0.2235
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.4-PO	2	0.9146+-0.0191	0.6927+-0.0040	0.7882+-0.0045
hg38_asm5_alt	dragmap-1.2.1	manta-1.6.0	2	0.9616+-0.0007	0.6550+-0.0169	0.7791+-0.0117
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.10	2	0.4718+-0.2763	0.6856+-0.0062	0.5085+-0.2036
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.10-PO	2	0.9013+-0.0195	0.6655+-0.0022	0.7655+-0.0056
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.4	2	0.4744+-0.2883	0.6665+-0.0052	0.5000+-0.2085
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.4-PO	2	0.9061+-0.0184	0.6463+-0.0002	0.7544+-0.0062
hg38_asm5_alt	sentieon-202112.01	manta-1.6.0	2	0.9563+-0.0018	0.6899+-0.0149	0.8015+-0.0094
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.10	2	0.4755+-0.2785	0.6851+-0.0058	0.5107+-0.2041
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.10-PO	2	0.8991+-0.0206	0.6651+-0.0028	0.7645+-0.0056
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.4	2	0.4791+-0.2912	0.6656+-0.0080	0.5016+-0.2078
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.4-PO	2	0.9059+-0.0176	0.6464+-0.0042	0.7543+-0.0032
hg38_asm5_alt	sentieon-202112.01-recal	manta-1.6.0	2	0.9547+-0.0018	0.6896+-0.0140	0.8007+-0.0088
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.10	2	0.6929+-0.0516	0.6802+-0.0128	0.6850+-0.0188
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.10-PO	2	0.9411+-0.0039	0.6406+-0.0104	0.7623+-0.0061
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.4	2	0.7062+-0.0531	0.6590+-0.0077	0.6805+-0.0207
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.4-PO	2	0.9430+-0.0048	0.6225+-0.0043	0.7500+-0.0016
hg38_asm5_alt	snap-2.0.0	manta-1.6.0	2	0.9509+-0.0022	0.6791+-0.0127	0.7923+-0.0079

6.1.2 Preparation: "PacBio-CCS-high"

Reference	Aligner	Caller	N	precision	recall	f1
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.10	1	0.9060+-0.0000	0.9799+-0.0000	0.9415+-0.0000
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.10-PO	1	0.9463+-0.0000	0.9729+-0.0000	0.9594+-0.0000
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.4	1	0.8962+-0.0000	0.9811+-0.0000	0.9367+-0.0000
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.4-PO	1	0.9415+-0.0000	0.9731+-0.0000	0.9571+-0.0000
hg38_T2T_masked	minimap2-2.23	pbsv-2.8.0	1	0.9463+-0.0000	0.9847+-0.0000	0.9651+-0.0000
hg38_T2T_masked	minimap2-2.23	sniffles-2.0.2	1	0.9494+-0.0000	0.9808+-0.0000	0.9649+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.10	1	0.8939+-0.0000	0.9803+-0.0000	0.9351+-0.0000

Reference	Aligner	Caller	N	precision	recall	f1
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.10-PO	1	0.9372+-0.0000	0.9729+-0.0000	0.9547+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.4	1	0.8869+-0.0000	0.9760+-0.0000	0.9293+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.4-PO	1	0.9554+-0.0000	0.1849+-0.0000	0.3098+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	pbsv-2.8.0	1	0.9420+-0.0000	0.9859+-0.0000	0.9634+-0.0000
hg38_T2T_masked	pbmm2-1.7.0	sniffles-2.0.2	1	0.9450+-0.0000	0.9811+-0.0000	0.9627+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.10	1	0.9079+-0.0000	0.9806+-0.0000	0.9428+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.10-PO	1	0.9468+-0.0000	0.9736+-0.0000	0.9600+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.4	1	0.8955+-0.0000	0.9806+-0.0000	0.9361+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.4-PO	1	0.9415+-0.0000	0.9727+-0.0000	0.9568+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	pbsv-2.8.0	1	0.9459+-0.0000	0.9847+-0.0000	0.9649+-0.0000
hg38_T2T_masked	sentieon_mm2-202112.01	sniffles-2.0.2	1	0.9492+-0.0000	0.9808+-0.0000	0.9647+-0.0000

6.2 Unrestricted calls

These evaluations are **UNRESTRICTED**, meaning that there is no BED file used to limit the analysis to a subset of the genome (e.g. no high-confidence region applied). As a result, precision results are skewed downwards and should be considered a lower-bound on precision with respect to the full benchmark dataset. However, this allows us to add benchmarks that were not released with a BED file, specifically HG001/NA12878's Personalis benchmark set.

6.2.1 Preparation: "PCR-free-illumina"

Reference	Aligner	Caller	N	precision	recall	f1
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.10	4	0.2677+-0.0975	0.8023+-0.1594	0.3864+-0.0979
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.10-PO	4	0.5638+-0.0896	0.7866+-0.1637	0.6354+-0.0077
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.4	4	0.2646+-0.0970	0.8008+-0.1597	0.3829+-0.0991
hg38_T2T_masked	dragmap-1.2.1	dysgu-1.3.4-PO	4	0.5682+-0.0907	0.7865+-0.1645	0.6380+-0.0072
hg38_T2T_masked	dragmap-1.2.1	manta-1.6.0	4	0.5691+-0.0968	0.7417+-0.1500	0.6221+-0.0078
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.10	4	0.2544+-0.0808	0.7661+-0.1549	0.3697+-0.0784
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.10-PO	4	0.5717+-0.0914	0.7482+-0.1599	0.6260+-0.0068
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.4	4	0.2468+-0.0789	0.7535+-0.1552	0.3599+-0.0782
hg38_T2T_masked	sentieon-202112.01	dysgu-1.3.4-PO	4	0.5693+-0.0916	0.7369+-0.1598	0.6199+-0.0078

Reference	Aligner	Caller	N	precision	recall	f1
hg38_T2T_masked	sentieon-202112.01	manta-1.6.0	4	0.5404+-0.0990	0.7589+-0.1444	0.6098+-0.0175
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.10	4	0.2569+-0.0810	0.7658+-0.1558	0.3725+-0.0783
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.10-PO	4	0.5714+-0.0919	0.7485+-0.1599	0.6258+-0.0064
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.4	4	0.2484+-0.0787	0.7525+-0.1552	0.3615+-0.0774
hg38_T2T_masked	sentieon-202112.01-recal	dysgu-1.3.4-PO	4	0.5692+-0.0924	0.7371+-0.1589	0.6198+-0.0064
hg38_T2T_masked	sentieon-202112.01-recal	manta-1.6.0	4	0.5392+-0.0979	0.7588+-0.1451	0.6092+-0.0167
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.10	4	0.3052+-0.0517	0.7535+-0.1597	0.4216+-0.0250
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.10-PO	4	0.6112+-0.0957	0.7082+-0.1522	0.6336+-0.0120
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.4	4	0.2768+-0.0463	0.7310+-0.1515	0.3903+-0.0248
hg38_T2T_masked	snap-2.0.0	dysgu-1.3.4-PO	4	0.6057+-0.0977	0.6900+-0.1455	0.6229+-0.0111
hg38_T2T_masked	snap-2.0.0	manta-1.6.0	4	0.4960+-0.0947	0.7504+-0.1473	0.5758+-0.0217
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.10	4	0.2825+-0.1061	0.7802+-0.1546	0.3981+-0.1023
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.10-PO	4	0.5743+-0.0933	0.7661+-0.1580	0.6348+-0.0078
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.4	4	0.2714+-0.1009	0.7783+-0.1542	0.3868+-0.1000
hg38_asm5_alt	dragmap-1.2.1	dysgu-1.3.4-PO	4	0.5745+-0.0922	0.7648+-0.1575	0.6346+-0.0062
hg38_asm5_alt	dragmap-1.2.1	manta-1.6.0	4	0.5851+-0.1027	0.7229+-0.1443	0.6242+-0.0073
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.10	4	0.2626+-0.0854	0.7642+-0.1553	0.3777+-0.0813
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.10-PO	4	0.5686+-0.0904	0.7475+-0.1594	0.6241+-0.0072
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.4	4	0.2164+-0.0636	0.7461+-0.1528	0.3261+-0.0658
hg38_asm5_alt	sentieon-202112.01	dysgu-1.3.4-PO	4	0.5629+-0.0909	0.7279+-0.1555	0.6130+-0.0078
hg38_asm5_alt	sentieon-202112.01	manta-1.6.0	4	0.5502+-0.1032	0.7558+-0.1430	0.6148+-0.0182
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.10	4	0.2646+-0.0856	0.7644+-0.1560	0.3799+-0.0813
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.10-PO	4	0.5677+-0.0898	0.7477+-0.1600	0.6235+-0.0067
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.4	4	0.2178+-0.0639	0.7458+-0.1533	0.3276+-0.0656
hg38_asm5_alt	sentieon-202112.01-recal	dysgu-1.3.4-PO	4	0.5628+-0.0919	0.7274+-0.1549	0.6126+-0.0057
hg38_asm5_alt	sentieon-202112.01-recal	manta-1.6.0	4	0.5492+-0.1020	0.7553+-0.1435	0.6141+-0.0173
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.10	4	0.3197+-0.0546	0.7492+-0.1579	0.4346+-0.0246
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.10-PO	4	0.6121+-0.0976	0.7053+-0.1509	0.6327+-0.0108
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.4	4	0.2781+-0.0471	0.7277+-0.1532	0.3908+-0.0252
hg38_asm5_alt	snap-2.0.0	dysgu-1.3.4-PO	4	0.6027+-0.0977	0.6880+-0.1475	0.6199+-0.0122
hg38_asm5_alt	snap-2.0.0	manta-1.6.0	4	0.4999+-0.0970	0.7479+-0.1467	0.5774+-0.0223

6.2.2   Preparation: "PacBio-CCS-high"

Reference	Aligner	Caller	N	precision	recall	f1
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.10	2	0.2805+-0.0825	0.9526+-0.0184	0.4259+-0.0970
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.10-PO	2	0.4273+-0.1364	0.9429+-0.0214	0.5732+-0.1263
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.4	2	0.2508+-0.0756	0.9544+-0.0156	0.3906+-0.0938
hg38_T2T_masked	minimap2-2.23	dysgu-1.3.4-PO	2	0.4148+-0.1336	0.9438+-0.0201	0.5618+-0.1264
hg38_T2T_masked	minimap2-2.23	pbsv-2.8.0	2	0.3681+-0.1158	0.9564+-0.0071	0.5204+-0.1206
hg38_T2T_masked	minimap2-2.23	sniffles-2.0.2	2	0.3764+-0.1182	0.9473+-0.0192	0.5265+-0.1189
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.10	2	0.2938+-0.0820	0.9559+-0.0166	0.4423+-0.0945
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.10-PO	2	0.4160+-0.1336	0.9440+-0.0172	0.5632+-0.1267
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.4	2	0.2589+-0.0754	0.9506+-0.0092	0.4008+-0.0926
hg38_T2T_masked	pbmm2-1.7.0	dysgu-1.3.4-PO	2	0.6125+-0.0607	0.4066+-0.2475	0.4280+-0.1706
hg38_T2T_masked	pbmm2-1.7.0	pbsv-2.8.0	2	0.3813+-0.1210	0.9593+-0.0087	0.5338+-0.1234
hg38_T2T_masked	pbmm2-1.7.0	sniffles-2.0.2	2	0.3836+-0.1205	0.9486+-0.0172	0.5339+-0.1203
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.10	2	0.2796+-0.0816	0.9532+-0.0186	0.4250+-0.0961
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.10-PO	2	0.4275+-0.1363	0.9434+-0.0216	0.5736+-0.1261
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.4	2	0.2497+-0.0748	0.9540+-0.0156	0.3893+-0.0929
hg38_T2T_masked	sentieon_mm2-202112.01	dysgu-1.3.4-PO	2	0.4148+-0.1338	0.9434+-0.0201	0.5617+-0.1265
hg38_T2T_masked	sentieon_mm2-202112.01	pbsv-2.8.0	2	0.3672+-0.1156	0.9561+-0.0070	0.5195+-0.1206
hg38_T2T_masked	sentieon_mm2-202112.01	sniffles-2.0.2	2	0.3761+-0.1180	0.9473+-0.0192	0.5262+-0.1187