Supplements:

Table S1. (a). Detailed neural network structural of UNet

Layer (type)	Output Shape	Param #	Connected to
input (InputLayer)	(None, 400, 1)	0	
conv1d_1 (Conv1D)	(None, 400, 32)	384	input[0][0]
batch_normalization_1 (BatchNor	(None, 400, 32)	128	conv1d_1[0][0]
conv1d_2 (Conv1D)	(None, 400, 32)	11296	batch_normalization_1[0][0]
batch_normalization_2 (BatchNor	(None, 400, 32)	128	conv1d_2[0][0]
max_pooling1d_1 (MaxPooling1D)	(None, 80, 32)	0	batch_normalization_2[0][0]
conv1d_3 (Conv1D)	(None, 80, 64)	22592	max_pooling1d_1[0][0]
batch_normalization_3 (BatchNor	(None, 80, 64)	256	conv1d_3[0][0]
conv1d_4 (Conv1D)	(None, 80, 64)	45120	batch_normalization_3[0][0]
batch_normalization_4 (BatchNor	(None, 80, 64)	256	conv1d_4[0][0]
max_pooling1d_2 (MaxPooling1D)	(None, 16, 64)	0	batch_normalization_4[0][0]
conv1d_5 (Conv1D)	(None, 16, 128)	90240	max_pooling1d_2[0][0]
batch_normalization_5 (BatchNor	(None, 16, 128)	512	conv1d_5[0][0]
conv1d_6 (Conv1D)	(None, 16, 128)	180352	batch_normalization_5[0][0]
batch_normalization_6 (BatchNor	(None, 16, 128)	512	conv1d_6[0][0]
max_pooling1d_3 (MaxPooling1D)	(None, 8, 128)	0	batch_normalization_6[0][0]
conv1d_7 (Conv1D)	(None, 8, 256)	360704	max_pooling1d_3[0][0]
batch_normalization_7 (BatchNor	(None, 8, 256)	1024	conv1d_7[0][0]
conv1d_8 (Conv1D)	(None, 8, 256)	721152	batch_normalization_7[0][0]
batch_normalization_8 (BatchNor	(None, 8, 256)	1024	conv1d_8[0][0]
up_sampling1d_1 (UpSampling1D)	(None, 16, 256)	0	batch_normalization_8[0][0]
concatenate_1 (Concatenate)	(None, 16, 384)	0	batch_normalization_6[0][0] up_sampling1d_1[0][0]
conv1d_9 (Conv1D)	(None, 16, 128)	540800	concatenate_1[0][0]
batch_normalization_9 (BatchNor	(None, 16, 128)	512	conv1d_9[0][0]
conv1d_10 (Conv1D)	(None, 16, 128)	180352	batch_normalization_9[0][0]
batch_normalization_10 (BatchNo	(None, 16, 128)	512	conv1d_10[0][0]
up_sampling1d_2 (UpSampling1D)	(None, 80, 128)	0	batch_normalization_10[0][0]
concatenate_2 (Concatenate)	(None, 80, 192)	0	batch_normalization_4[0][0] up_sampling1d_2[0][0]
conv1d_11 (Conv1D)	(None, 80, 64)	135232	concatenate_2[0][0]
batch_normalization_11 (BatchNo	(None, 80, 64)	256	conv1d_11[0][0]
conv1d_12 (Conv1D)	(None, 80, 64)	45120	batch_normalization_11[0][0]
batch_normalization_12 (BatchNo	(None, 80, 64)	256	conv1d_12[0][0]
up_sampling1d_3 (UpSampling1D)	(None, 400, 64)	0	batch_normalization_12[0][0]
concatenate_3 (Concatenate)	(None, 400, 96)	0	batch_normalization_2[0][0] up_sampling1d_3[0][0]
conv1d_13 (Conv1D)	(None, 400, 32)	33824	concatenate_3[0][0]
batch_normalization_13 (BatchNo	(None, 400, 32)	128	conv1d_13[0][0]
conv1d_14 (Conv1D)	(None, 400, 32)	11296	batch_normalization_13[0][0]
batch_normalization_14 (BatchNo	(None, 400, 32)	128	conv1d_14[0][0]
conv1d_15 (Conv1D)	(None, 400, 2)	706	batch_normalization_14[0][0]
batch_normalization_15 (BatchNo	(None, 400, 2)	8	conv1d_15[0][0]
conv1d_16 (Conv1D)	(None, 400, 1)	3	batch_normalization_15[0][0]

Total params: 2,384,813
Trainable params: 2,381,993
Non-trainable params: 2,820

Table S1. (b). Detailed neural network structural of CNN

Layer (type)	Output Shape	Param #
conv1d_1 (Conv1D)	(None, 390, 64)	768
max_pooling1d_1 (MaxPooling1	(None, 78, 64)	0
conv1d_2 (Conv1D)	(None, 72, 128)	57472
max_pooling1d_2 (MaxPooling1	(None, 14, 128)	0
flatten_1 (Flatten)	(None, 1792)	0
dense_1 (Dense)	(None, 256)	459008
dropout_1 (Dropout)	(None, 256)	0
dense_2 (Dense)	(None, 400)	102800

Total params: 620,048
Trainable params: 620,048
Non-trainable params: 0

Table S2. (a).VCF files used in the evaluation

Samples	VCF files
Simulation	https://github.com/stat-lab/EvalSVcallers/blob/master/Ref_SV/Sim-A.SV.vcf
NA12878, NA19238, NA19239	ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/phase3/integrated_sv_map/ALL.wgs.mergedSV.v8.20130502.svs.genotypes.vcf.gz
HG002	ftp://ftp-trace.ncbi.nlm.nih.gov/giab/ftp/data/AshkenazimTrio/analysis/NIST_SVs_Integration_v0.6/HG002_SVs_Tier1_v0.6.vcf.gz

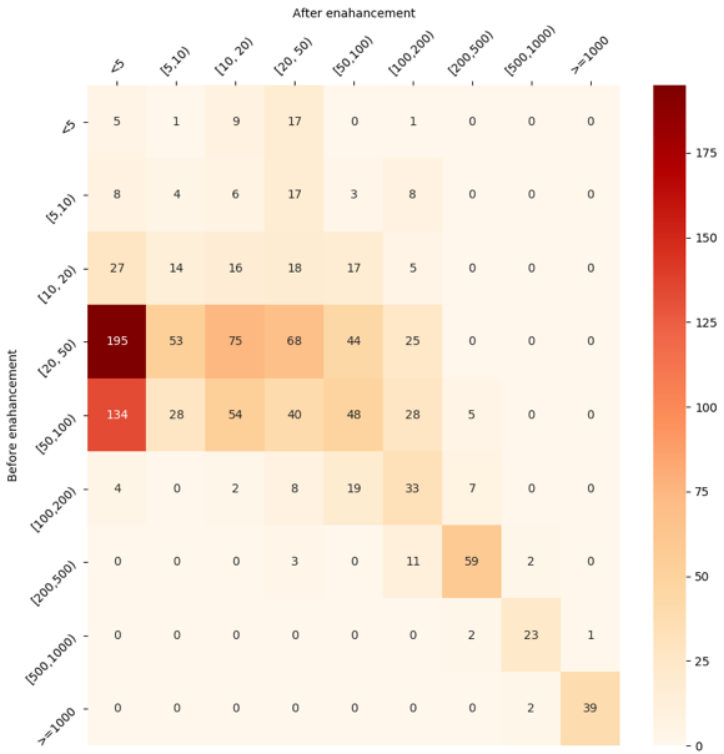
(b). BAM files

Samples	BAM files
NA12878	ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data/NA12878/high_coverage_alignment/NA12878.mapped.ILLUMINA.bwa.CEU.high_coverage_pcr_free.20130906.bam
NA19238	ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data/NA12878/high_coverage_alignment/NA19238.mapped.ILLUMINA.bwa.YRI.high_coverage_pcr_free.20130924.bam
NA19239	ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data/NA12878/high_coverage_alignment/NA19239.mapped.ILLUMINA.bwa.YRI.high_coverage_pcr_free.20130924.bam
HG002	ftp://ftp-trace.ncbi.nlm.nih.gov/giab/ftp/data/AshkenazimTrio/HG002_NA24385_son/NIST_HiSeq_HG002_Homogeneity-10953946/NHGRI_Illumina300X_AJtrio_novoalign_bams/HG002.hs37d5.60X.1.bam.

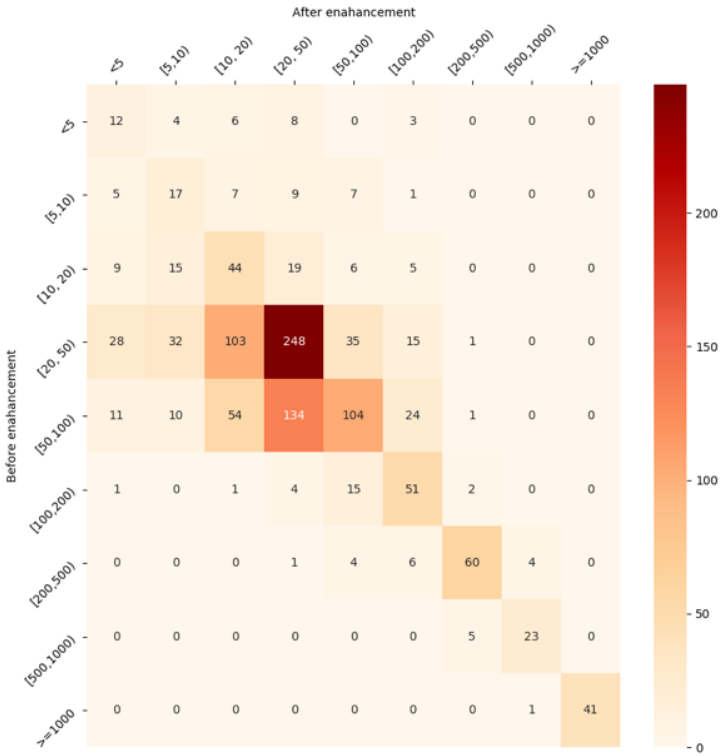
Figure S1. Breakpoint change matrix of in-sample enhancement.

NA12878

UNet

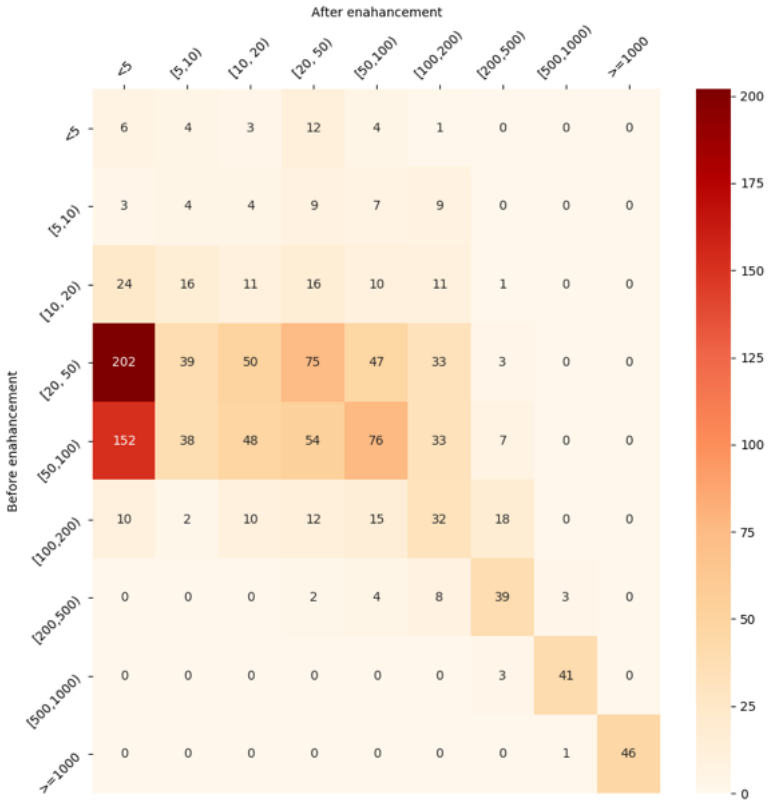


CNN

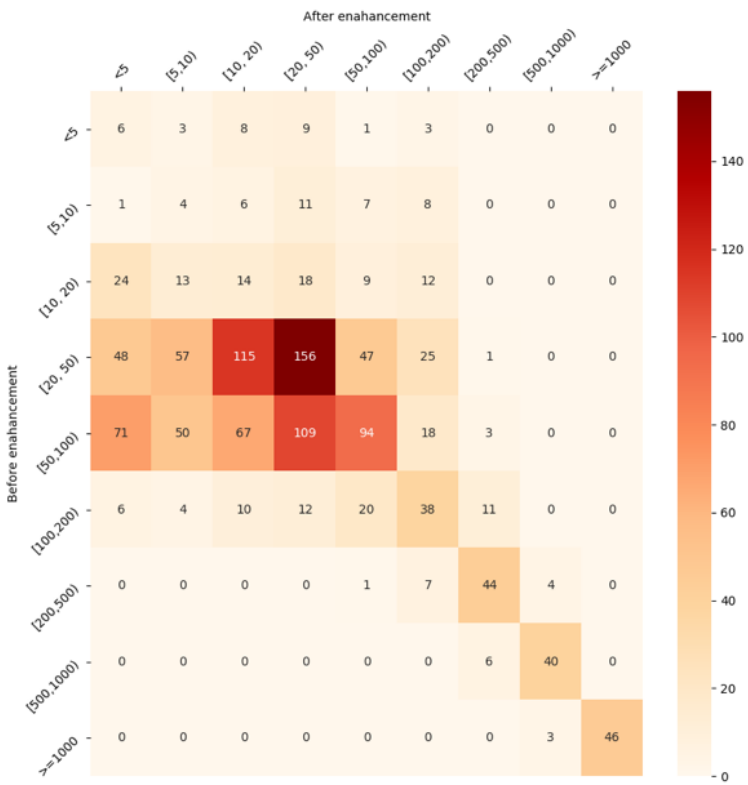


NA19238

UNet

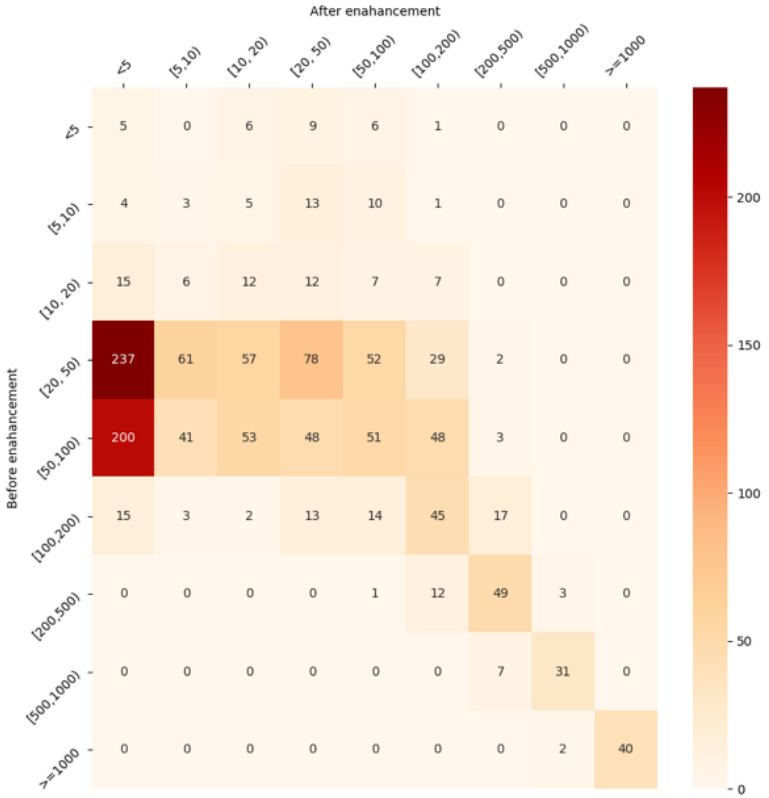


CNN

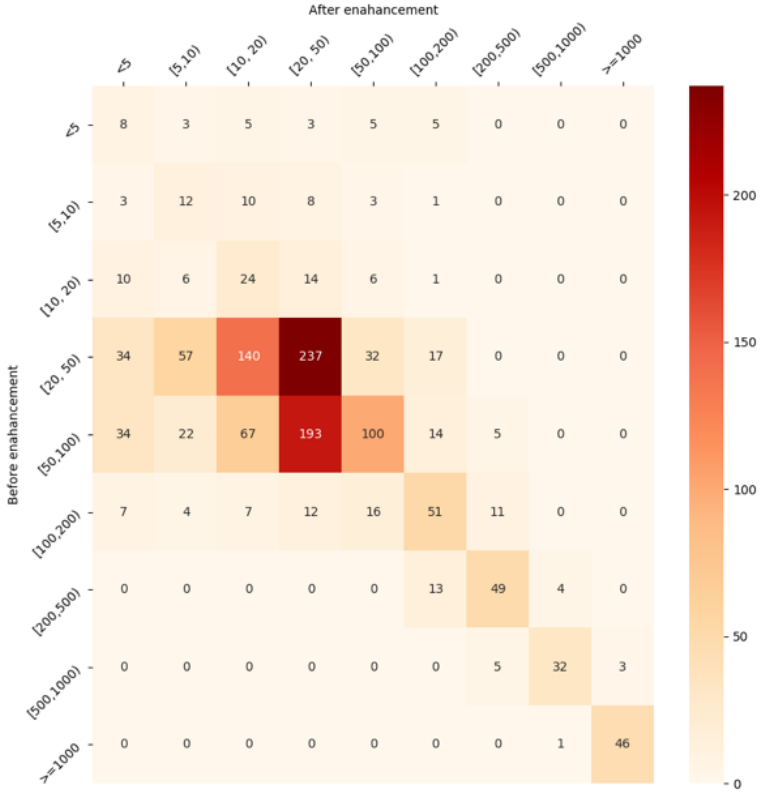


NA19239

UNet

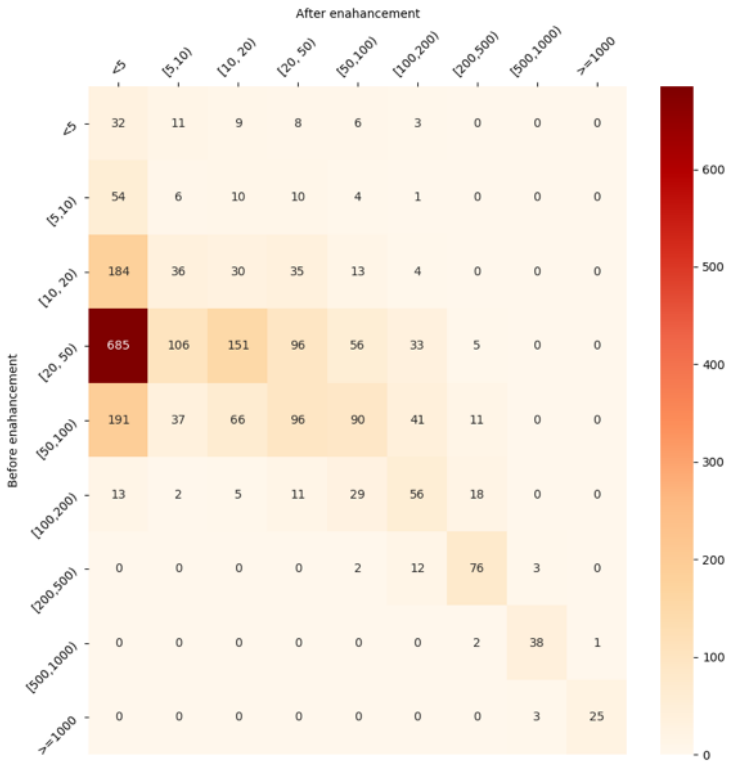


CNN



HG002

UNet



CNN

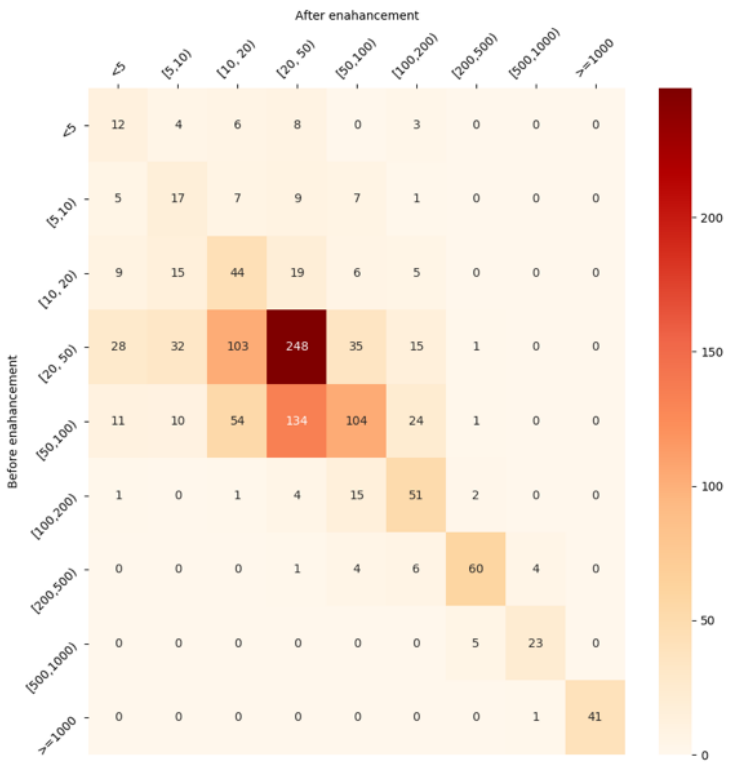
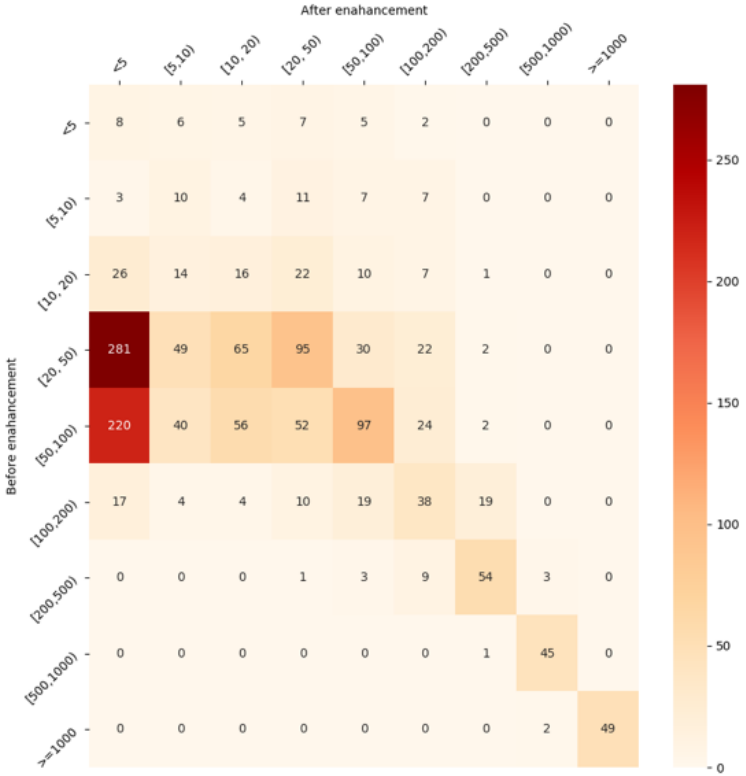


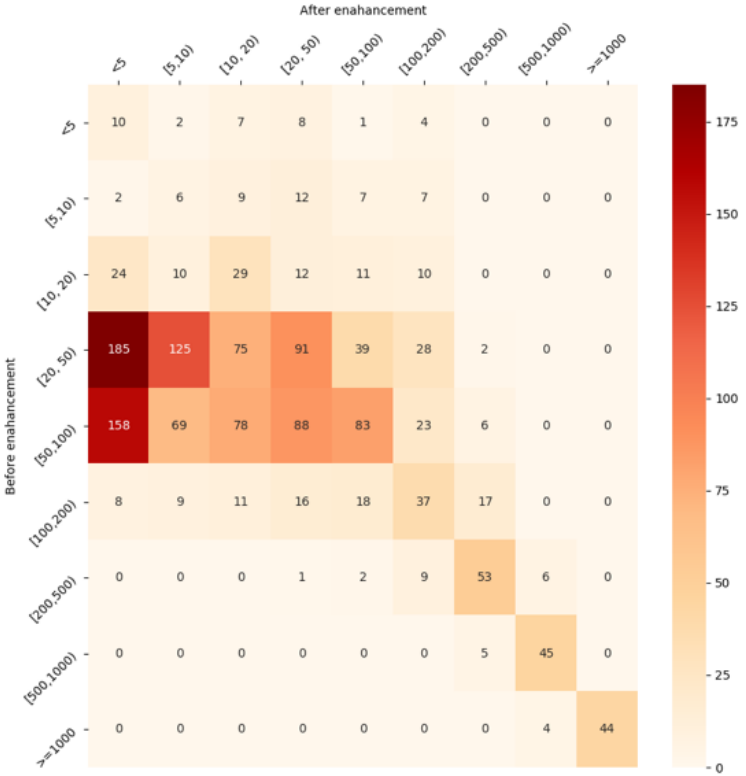
Figure S2. Breakpoint change matrix of cross-sample enhancement.

NA19238

UNet

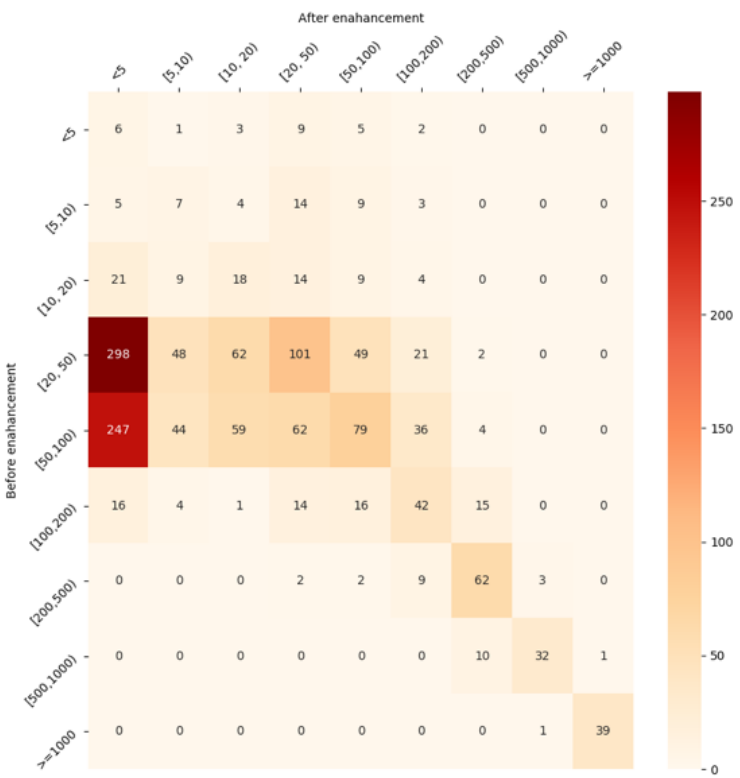


CNN



NA19239

UNet



CNN

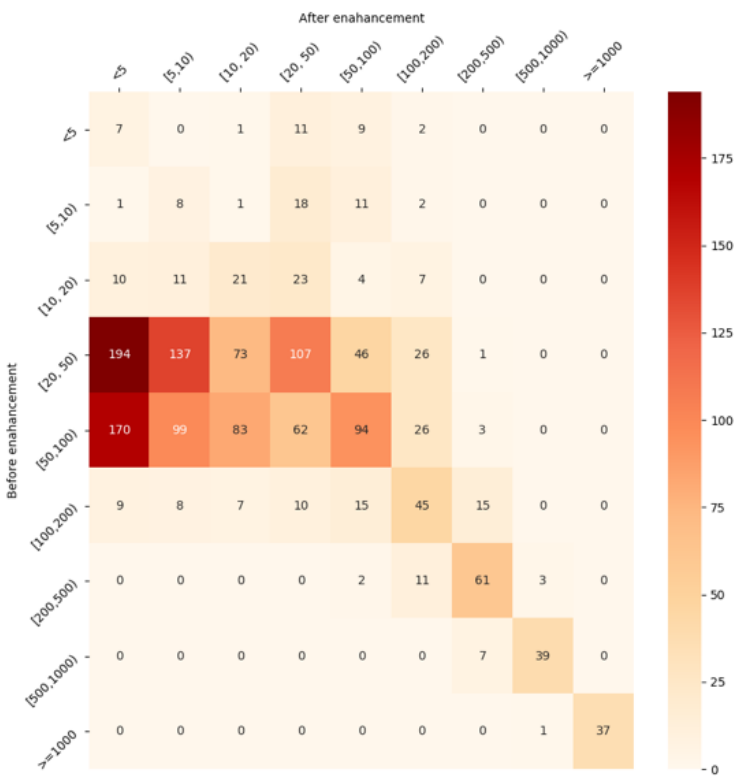
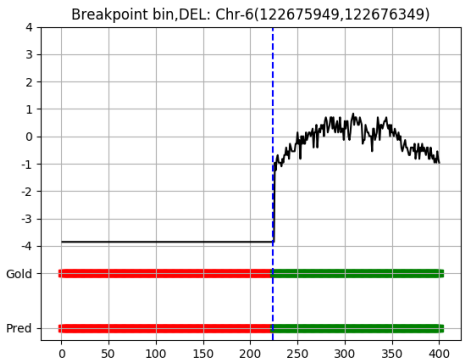


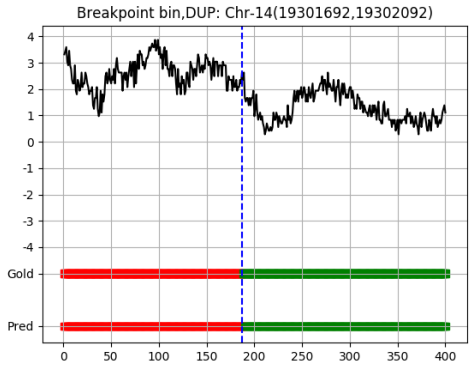
Figure S3. Segmentation examples on Simulation the data. Dash line indicates the position of the gold breakpoint. Coordinates in red are the SV related masks.

(a). Examples of positive enhancement by UNet

obvious case

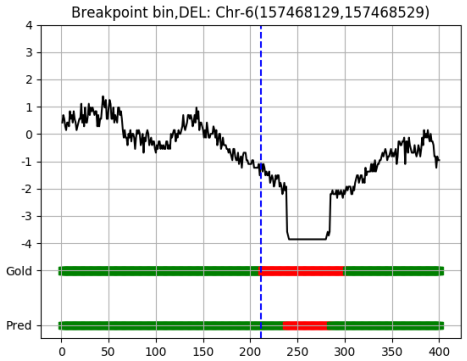


less obvious case

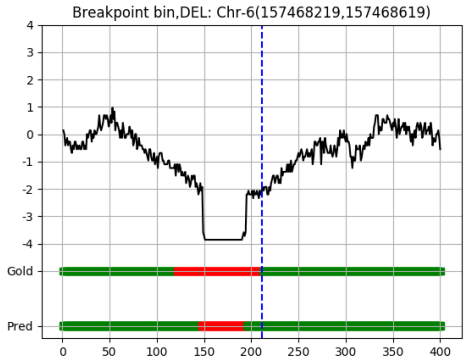


(b). small SVs totally inside the screening window

Unet

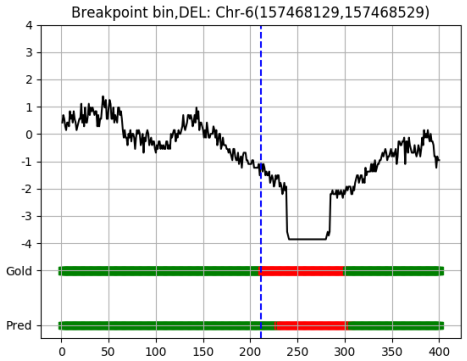


Left

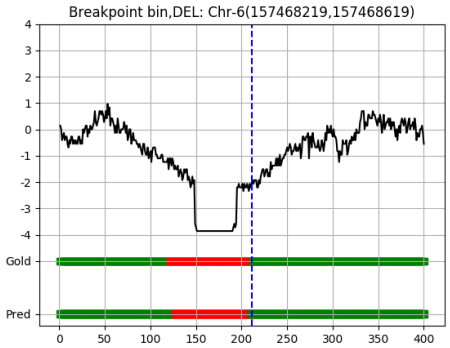


Right

CNN

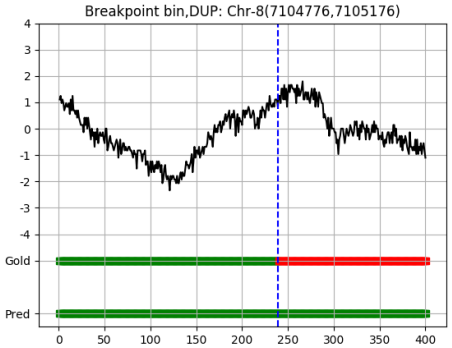


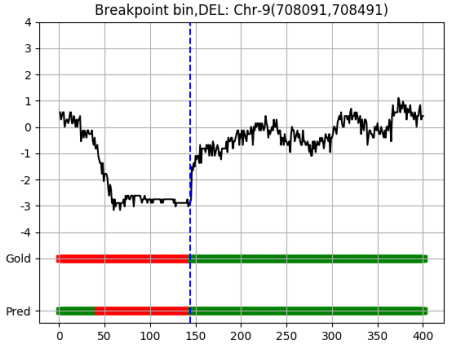
Left



Right

(c). Negative segmentation of UNet





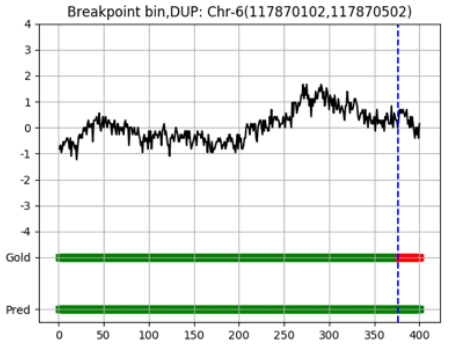


Table S3: The mean and standard derivation of to-gold-distances of different ranges for real data in-sample evaluation

		mean to-gold-distance										std to-gold-distance								
		<5	[5,10)	[10,20)	[20,50)	[50,100)	[100,200)	[200,500)	[500,1000)	>=1000		<5	[5,10)	[10,20)	[20,50)	[50,100)	[100,200)	[200,500)	[500,1000)	>=1000
na12878	/	2.61	7.17	14.38	34.84	64.26	138.01	315.54	670.18	7364.78		1.25	1.27	2.78	8.39	11.18	27.46	84.24	128.24	14794.04
	CNN	2.32	7.22	14.42	31.43	68.3	135.27	323.1	673.54	7250.29		1.32	1.31	2.81	8.27	14.08	27.92	88.19	138.21	14643.51
	Unet	1.42	6.95	13.99	32.44	70.69	140.38	314.6	668.66	7383.95		1.26	1.39	2.76	9.15	14.68	27.13	83.37	143.55	14782.42
na19238	/	2.5	7.14	14.94	35.43	65.46	141.07	331.91	684.02	4738.9		1.36	1.32	2.81	8.44	11.58	30.53	100.65	137.68	8350.85
	CNN	2.42	7.08	14.08	32.54	69.63	139.36	318.67	703.58	4928.44		1.27	1.41	2.87	8.54	13.14	28.21	91.73	146.83	8568.06
	Unet	1.39	6.73	14.14	32.07	71.84	140.89	302.6	690.13	4638.67		1.18	1.41	2.73	8.69	14.37	28.95	89.61	146.9	8289.47
na19239	/	2	7.32	14.87	35.6	64.81	140.02	308.49	696	3592.76		1.2	1.38	2.71	8.39	12.04	31.16	89.6	147.07	6264.21
	CNN	2.16	6.97	14.44	30.98	67.77	149.08	307.25	691.74	3498.3		1.21	1.42	2.8	7.98	13.98	31.28	81.62	142.4	6172.19
	Unet	1.11	6.82	13.77	33.29	71.81	140.13	306.04	728.76	3690.94		1.31	1.41	2.64	8.42	13.52	28.75	86.73	150.23	6363.64
HG002	/	2.12	7.2	14.55	34.1	62.53	143.1	302.61	759.22	2026.86		1.38	1.37	2.88	8.51	11.58	28.34	77.88	128.36	1978.93
	CNN	2.1	6.88	14.14	30.98	67.3	142.33	295.96	767.19	2100.7		1.33	1.44	2.87	8.35	13.42	28.08	77.41	126.57	2021.22
	Unet	0.85	6.73	13.49	33.19	68.85	143.94	287.65	753.48	2136.5		1.29	1.48	2.62	9.14	13.06	29.82	74.57	126.1	2038.64

Table S4: derivation of to-gold-distances of different ranges for real data cross-sample evaluation

		mean to-gold-distance										std to-gold-distance								
		<5	[5,10)	[10,20)	[20,50)	[50,100)	[100,200)	[200,500)	[500,1000)	>=1000		<5	[5,10)	[10,20)	[20,50)	[50,100)	[100,200)	[200,500)	[500,1000)	>=1000
NA19238	/	2.35	7.27	15	35.17	65.55	141.26	329.71	689.41	4553.32		1.39	1.3	2.8	8.27	11.61	30.25	97.58	136.66	7581.48
	CNN	2.04	6.66	13.85	32.45	71.26	140.74	314.26	705.37	4709.07		1.24	1.4	2.86	8.28	14.26	28.69	90.31	148.2	7933.48
	Unet	1.29	6.62	13.84	32.44	70.82	143.58	310.9	690.37	4493.61		1.35	1.43	2.79	8.64	13.72	30.29	89.01	140.73	7707.92
NA19239	/	2.06	7.39	15.17	35.65	65.1	140.46	321.27	686.16	3923.15		1.25	1.37	2.76	8.41	11.96	31.29	91.95	141.3	6207.5
	CNN	2.13	6.69	13.6	31.83	70.95	141.58	322.54	705.53	3974.33		1.29	1.39	2.72	8.57	13.97	28.4	96.15	145.04	6249.28
	Unet	1.26	7.03	13.86	33.26	70.6	141.97	322.93	708.59	3932.57		1.33	1.44	2.71	8.94	12.88	30.1	92.15	128.35	6207.13

Table S5: Each repeat one result of 5-fold cross validation on the simulation data.

80% train 20% test										
	RunID	AUC	Sensitivity	FDR	Precision	Recall	All-dice	BK-dice	all-IOU	BK-IOU
Unet	1	0.8911	0.8619	0.0837	0.9163	0.8619	0.822	0.8503	0.6979	0.7397
	2	0.8945	0.8588	0.0741	0.9259	0.8588	0.8231	0.8489	0.6995	0.7376
	3	0.8951	0.8544	0.069	0.931	0.8544	0.8268	0.8487	0.7048	0.7372
	4	0.8957	0.8465	0.0604	0.9396	0.8465	0.8247	0.843	0.7018	0.7286
	5	0.8936	0.8513	0.0689	0.9311	0.8513	0.8232	0.8455	0.6996	0.7324
	AVG	0.894	0.85458	0.07122	0.92878	0.85458	0.82396	0.84728	0.70072	0.7351
SVM		0.8661	0.8133	0.0898	0.9102	0.8133				
CNN	1	0.8747	0.8428	0.0988	0.9012	0.8428	0.7957	0.8238	0.6607	0.7004
	2	0.8788	0.8531	0.0996	0.9004	0.8531	0.801	0.8286	0.6681	0.7074
	3	0.8751	0.859	0.111	0.889	0.859	0.7986	0.8293	0.6647	0.7083
	4	0.8751	0.8519	0.1055	0.8945	0.8519	0.7983	0.8281	0.6643	0.7066
	5	0.8758	0.8523	0.1042	0.8958	0.8523	0.7957	0.8252	0.6608	0.7025
	AVG	0.8759	0.85182	0.10382	0.89618	0.85182	0.79786	0.827	0.66372	0.70504

20% train 80% test										
		AUC	Sensitivity	FDR	Precision	Recall	All-dice	BK-dice	all-IOU	BK-IOU
Unet	1	0.8817	0.8514	0.0922	0.9078	0.8514	0.7974	0.8247	0.6631	0.7019
	2	0.8844	0.8511	0.0866	0.9134	0.8511	0.8066	0.8343	0.676	0.7157
	3	0.8831	0.8476	0.0861	0.9139	0.8476	0.8033	0.8337	0.6712	0.7149
	4	0.8802	0.8336	0.078	0.922	0.8336	0.8037	0.8271	0.6719	0.7054
	5	0.883	0.8594	0.0967	0.9033	0.8594	0.8042	0.8356	0.6726	0.7178
	AVG	0.88248	0.84862	0.08792	0.91208	0.84862	0.80304	0.83108	0.67096	0.71114
SVM		0.8576	0.8057	0.0998	0.9002	0.8057				
CNN		0.8432	0.8223	0.1394	0.8606	0.8223	0.7476	0.7853	0.5971	0.6467
		0.8394	0.8363	0.1567	0.8433	0.8363	0.7469	0.7903	0.5964	0.6536
		0.8438	0.8334	0.1461	0.8539	0.8334	0.7504	0.7925	0.6005	0.6565
		0.8424	0.8172	0.1378	0.8622	0.8172	0.7454	0.7848	0.5942	0.646
		0.8445	0.8237	0.1391	0.8609	0.8237	0.7489	0.7883	0.5987	0.6507
	AVG	0.84266	0.82658	0.14382	0.85618	0.82658	0.74784	0.78824	0.59738	0.6507