

11th May 2024

Detection of Synthetic CNVs using CuteSV, Sniffles2 and SVIM

Homozygous Deletions

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	30	35	49	49	48
300-500	29	38	51	51	48
501-1000	23	36	49	49	47
1500-2000	25	36	50	50	49
4000-5000	25	39	50	50	49
8000-10000	26	37	50	50	49
25000-30000	29	38	50	50	50

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	50	46	50	50	50
300-500	51	50	51	51	51
501-1000	49	48	49	49	49
1500-2000	49	46	50	50	50
4000-5000	50	48	50	50	50
8000-10000	50	47	50	50	50
25000-30000	0	0	0	0	0

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	50	49	49	50	50
300-500	51	51	51	51	51
501-1000	49	48	49	49	49
1500-2000	50	48	51	50	50
4000-5000	50	49	50	50	50
8000-10000	50	48	50	51	50
25000-30000	50	49	50	50	50

Heterozygous Deletions

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	1	24	40	39	44
300-500	1	27	43	41	44
501-1000	0	22	44	39	45
1500-2000	0	25	45	41	45
4000-5000	1	19	44	41	45
8000-10000	0	28	44	42	45
25000-30000	0	22	44	39	45

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	50	50	45	45	45
300-500	50	51	45	46	45
501-1000	48	49	46	44	45
1500-2000	48	49	45	45	45
4000-5000	46	50	45	45	45
8000-10000	48	49	45	45	46
25000-30000	47	50	45	45	45

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	50	49	45	46	45
300-500	50	51	45	46	47
501-1000	49	49	46	44	47
1500-2000	49	50	45	46	46
4000-5000	50	50	46	45	45
8000-10000	50	50	46	47	46
25000-30000	50	50	46	46	47

Homozygous Duplications with Copy Number 3

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	19	24	30	26	27
300-500	7	24	31	29	34
501-1000	7	23	31	27	31
1500-2000	7	19	40	41	52
4000-5000	0	18	42	31	46
8000-10000	11	23	30	26	27
25000-30000	13	23	30	26	27

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	30	27	30	30	27
300-500	40	32	31	30	28
501-1000	36	42	37	41	35
1500-2000	41	60	82	67	60
4000-5000	47	42	72	70	75
8000-10000	29	27	30	31	27
25000-30000	29	27	30	29	28

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	30	28	30	31	27
300-500	35	35	42	40	43
501-1000	59	69	79	76	76
1500-2000	112	103	142	128	134
4000-5000	87	105	183	163	186
8000-10000	30	29	31	35	29
25000-30000	30	29	30	29	30

Heterozygous Duplications with Copy Number 3

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	0	15	27	22	21
300-500	0	14	24	24	21
501-1000	1	12	26	21	21
1500-2000	0	3	25	29	26
4000-5000	0	3	15	23	28
8000-10000	0	8	26	23	21
25000-30000	0	14	27	25	21

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	27	28	29	29	24
300-500	26	29	31	30	24
501-1000	24	30	37	30	26
1500-2000	13	25	29	34	37
4000-5000	31	33	41	41	40
8000-10000	26	27	28	30	22
25000-30000	28	30	27	30	23

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	30	30	29	30	28
300-500	34	36	36	37	33
501-1000	56	69	69	72	65
1500-2000	74	108	119	128	111
4000-5000	58	74	109	118	127
8000-10000	30	30	30	31	26
25000-30000	30	30	29	30	26

Homozygous Duplications with Copy Number 4

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	22	31	39	36	38
300-500	18	31	36	38	46
501-1000	15	28	35	43	43
1500-2000	7	27	50	68	65
4000-5000	5	24	41	55	64
8000-10000	10	34	37	36	36
25000-30000	17	33	38	36	37

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	40	40	40	36	40
300-500	53	46	44	36	42
501-1000	63	62	58	44	52
1500-2000	73	85	101	90	88
4000-5000	70	82	75	91	95
8000-10000	40	40	40	36	40
25000-30000	40	40	40	36	40

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	40	40	40	36	41
300-500	44	48	56	61	63
501-1000	87	99	109	106	106
1500-2000	148	166	176	168	181
4000-5000	130	161	215	258	263
8000-10000	39	40	40	36	40
25000-30000	39	39	39	36	40

Heterozygous Duplications with Copy Number 4

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	2	19	35	38	40
300-500	0	15	35	38	42
501-1000	0	17	35	37	39
1500-2000	0	7	29	40	52
4000-5000	0	2	28	33	40
8000-10000	2	15	34	39	38
25000-30000	0	21	35	37	37

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	37	39	38	40	40
300-500	39	50	42	45	44
501-1000	24	49	56	46	48
1500-2000	20	38	42	59	68
4000-5000	36	49	58	65	64
8000-10000	35	39	40	40	40
25000-30000	37	39	40	40	40

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	40	40	40	40	40
300-500	46	48	49	49	58
501-1000	73	87	105	105	113
1500-2000	108	144	161	175	177
4000-5000	89	117	174	186	216
8000-10000	36	40	40	40	40
25000-30000	37	40	40	40	40

Homozygous Duplications with Copy Number 5

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	30	44	46	46	45
300-500	25	49	51	60	56
501-1000	14	48	49	50	50
1500-2000	6	42	67	91	86
4000-5000	3	36	55	76	85
8000-10000	22	49	50	50	45
25000-30000	24	49	50	50	45

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	50	50	50	50	48
300-500	57	54	50	50	51
501-1000	83	76	67	55	56
1500-2000	85	112	134	123	108
4000-5000	76	102	111	131	132
8000-10000	49	50	50	50	50
25000-30000	45	46	46	46	46

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	47	46	47	46	48
300-500	57	59	67	72	78
501-1000	113	125	133	133	136
1500-2000	196	213	234	232	227
4000-5000	153	216	284	337	346
8000-10000	50	50	51	52	51
25000-30000	50	50	50	50	50

Heterozygous Duplications with Copy Number 5

CuteSV_summary

Bin	5x	10x	20x	30x	40x
50-100	6	29	46	41	33
300-500	3	29	49	44	39
501-1000	1	17	50	45	40
1500-2000	0	14	45	47	47
4000-5000	0	3	33	42	37
8000-10000	0	19	48	44	36
25000-30000	3	20	50	44	38

Sniffles_summary

Bin	5x	10x	20x	30x	40x
50-100	42	50	50	49	47
300-500	40	50	50	47	46
501-1000	47	63	60	57	51
1500-2000	28	47	70	66	58
4000-5000	51	70	78	72	75
8000-10000	47	49	50	48	48
25000-30000	43	46	46	44	42

SVIM_summary

Bin	5x	10x	20x	30x	40x
50-100	43	46	47	46	43
300-500	53	62	58	60	57
501-1000	88	101	123	125	114
1500-2000	136	188	216	206	200
4000-5000	120	141	219	234	228
8000-10000	48	50	51	48	49
25000-30000	50	50	50	49	47