

Report CORIANDR: ChrOmosomal abeRration Identifier AND Reporter in R

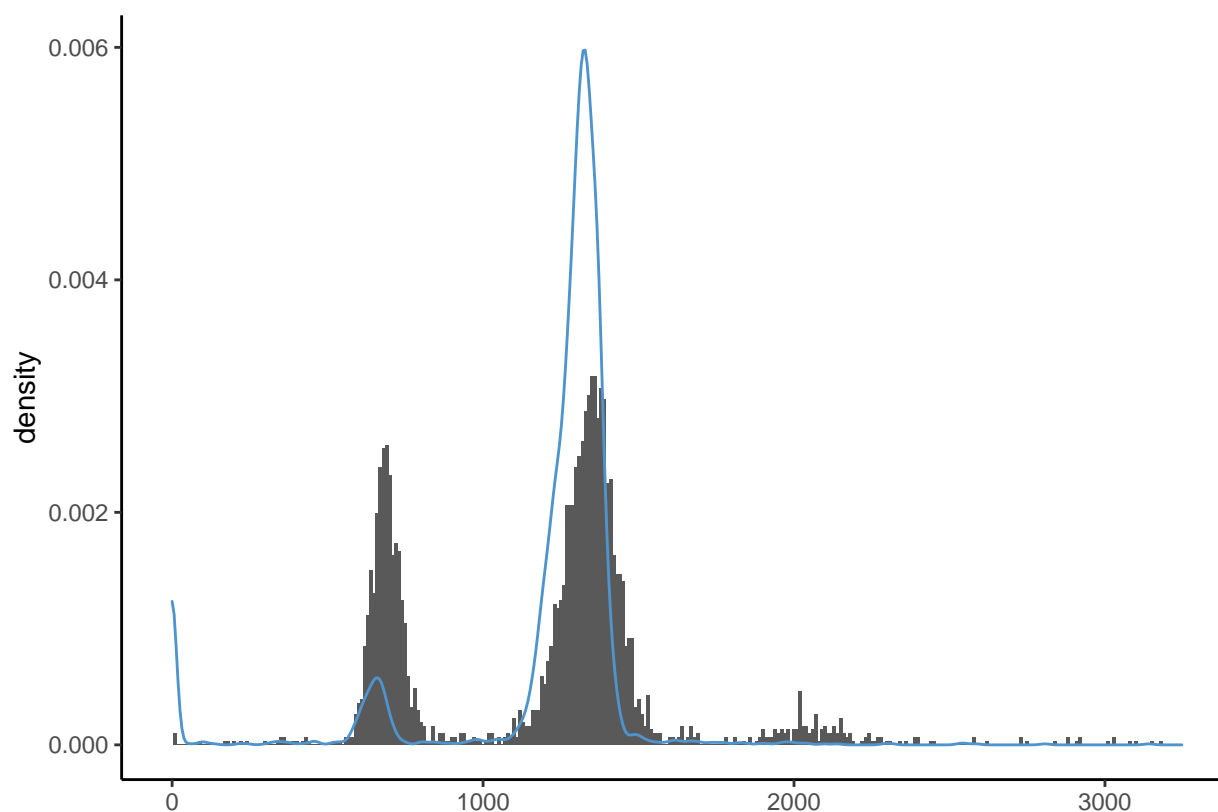
18 März, 2021

Sample ID

120517.new.report

row read pairs	1922410
average read length	149.5
unique mapped pairs	1846510
gender	M

Distribution of bin depth in sample



This graph shows the distribution of the bins according to the depth of the sequencing. The blue curve represents the distribution of the bins in the PON. Ideally, the histogram and the blue curve match completely. If not, this can indicate larger deletions (the first peak decreases) or larger amplifications (the third peak increases).

Calculated numerical karyotype:

add(1)(p36.33p36.33)
del(1)(p36.21p36.21)
del(1)(p36.12p36.11)
del(1)(p34.2p34.2)
add(1)(p33p33)
add(1)(p31.2p31.2)
add(1)(p22.2p22.2)
add(1)(p21.3p21.3)
add(1)(q32.2q32.2)
add(1)(q42.13q42.3)
add(2)(p25.3p25.3)
add(2)(p25.1p24.3)
add(2)(p24.1p24.1)
del(2)(p23.3p23.3)
del(2)(p22.1p22.1)
add(2)(p16.3p16.3)
add(2)(p11.2p11.2)
del(2)(q11.1q37.3)
add(3)(p26.3p26.2)
add(3)(p25.1p24.3)
add(3)(p22.3p22.3)
del(3)(p21.31p21.31)
add(3)(p14.1p14.1)
add(3)(p12.3p12.2)
add(3)(p11.1q11.1)
add(3)(q12.1q12.1)
add(3)(q13.13q13.2)
add(3)(q13.32q21.1)
del(3)(q22.1q22.3)
add(3)(q24q24)
add(3)(q25.32q25.32)
add(3)(q26.1q26.1)
add(3)(q26.32q26.32)
add(4)(p16.3p16.3)
add(4)(p15.31p15.31)

add(4)(p13p11)
 add(4)(q12q12)
 add(4)(q21.23q21.23)
 add(4)(q22.1q22.1)
 add(4)(q23q25)
 add(4)(q27q27)
 add(4)(q28.2q28.2)
 add(4)(q31.3q31.3)
 add(4)(q35.2q35.2)
 add(5)(p15.33p15.33)
 add(5)(p15.2p15.2)
 add(5)(p13.3p13.3)
 add(5)(p12p12)
 add(5)(q11.1q12.1)
 add(5)(q12.3q12.3)
 del(5)(q15q23.1)
 del(5)(q23.3q31.1)
 add(5)(q31.3q32)
 del(5)(q33.1q33.3)
 add(5)(q34q35.1)
 del(5)(q35.2q35.2)
 add(6)(p25.3p25.3)
 del(6)(p21.33p21.33)
 del(6)(p21.31p21.2)
 del(6)(p11.2p11.2)
 del(6)(q13q27)
 add(7)(p12.3p12.1)
 add(7)(q11.21q11.21)
 del(7)(q11.23q11.23)
 add(7)(q21.11q21.12)
 add(7)(q21.3q21.3)
 add(7)(q22.2q22.2)
 add(7)(q31.32q31.32)
 del(7)(q32.3q32.3)
 add(7)(q36.2q36.2)
 del(8)(p23.3p11.22)

add(8)(p11.21p11.21)
 add(8)(q11.1q24.3)
 add(9)(p24.3p24.3)
 add(9)(p22.3p22.3)
 add(9)(p21.3p21.2)
 add(9)(p13.2p13.2)
 add(9)(q31.1q31.2)
 add(9)(q32q32)
 del(9)(q33.3q34.3)
 add(10)(p15.3p15.3)
 del(10)(p14p14)
 add(10)(p11.1p11.1)
 add(10)(q11.23q21.1)
 add(10)(q21.3q21.3)
 del(10)(q22.1q22.1)
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 add(10)(q24.33q25.1)
 add(10)(q26.11q26.12)
 add(10)(q26.3q26.3)
 del(11)(p15.5p13)
 add(11)(p12p12)
 add(11)(p11.12q11)
 add(11)(q14.3q14.3)
 add(11)(q22.3q22.3)
 add(11)(q24.2q24.2)
 add(11)(q25q25)
 del(12)(p13.31p11.21)
 add(12)(q11q12)
 del(12)(q13.12q13.12)
 add(12)(q21.2q21.2)
 add(12)(q21.32q21.33)
 del(12)(q24.11q24.12)
 add(12)(q24.33q24.33)
 add(13)(p11.1q12.3)
 del(13)(q13.1q34)
 add(14)(p11.1q11.1)

add(14)(q12q12)
 add(14)(q13.3q13.3)
 add(14)(q21.2q21.2)
 del(14)(q24.2q24.2)
 add(14)(q31.2q31.3)
 del(14)(q32.13q32.13)
 add(15)(q13.1q13.1)
 add(15)(q14q14)
 del(15)(q15.1q15.1)
 del(15)(q21.2q21.2)
 del(15)(q24.1q24.3)
 add(15)(q25.1q25.2)
 add(15)(q26.3q26.3)
 del(16)(p13.11p12.3)
 del(16)(p11.2p11.1)
 del(16)(q11.2q24.3)
 del(17)(p13.3p12)
 add(17)(p11.2p11.2)
 del(17)(q12q12)
 del(17)(q21.32q21.32)
 del(17)(q22q22)
 del(18)(p11.31p11.1)
 del(18)(q11.2q23)
 del(19)(p13.3p13.2)
 del(19)(p13.12p13.11)
 del(19)(q13.2q13.2)
 del(19)(q13.32q13.42)
 del(20)(p13p11.1)
 add(20)(q11.1q11.22)
 del(20)(q11.23q13.33)
 add(21)(p12p11.1)
 add(21)(q21.1q21.1)
 add(21)(q21.3q21.3)
 add(21)(q22.12q22.12)
 add(22)(p11.2q12.2)
 del(22)(q12.3q13.1)

add(X)(p21.1p21.1)
add(X)(p11.21p11.21)
add(X)(q21.2q21.2)
add(X)(q21.32q21.32)
add(X)(q23q24)
add(X)(q26.1q26.1)
add(X)(q28q28)
add(Y)(p11.31p11.1)
add(Y)(q11.23q11.23)

Genes affected by CNVs*:

id	sym	bol chr	sta	rt end	abe	rration
882	ENSG00000117713	ARID1A	chr1	26693236	26782104	del
1258	ENSG00000127603	MACF1	chr1	39081316	39487177	del
1604	ENSG00000123080	CDKN2C	chr1	50960745	50974634	del
4895	ENSG00000163041	H3F3A	chr1	226061851	226072007	del
5713	ENSG00000134323	MYCN	chr2	15940550	15947007	amp
6626	ENSG00000169564	PCBP1	chr2	70087454	70089203	amp
7505	ENSG00000153094	BCL2L1	chr2	111119378	111168445	del
8082	ENSG00000121989	ACVR2A	chr2	147844517	147930826	del
8195	ENSG00000115170	ACVR1	chr2	157736444	157875862	del
8499	ENSG00000116044	NFE2L2	chr2	177227595	177392697	del
8662	ENSG00000064933	PMS1	chr2	189784085	189877629	del
8751	ENSG00000115524	SF3B1	chr2	197388515	197435091	del
8828	ENSG00000064012	CASP8	chr2	201233443	201287711	del
9003	ENSG00000138413	IDH1	chr2	208236227	208266074	del
9038	ENSG00000178568	ERBB4	chr2	211375717	212538841	del
9287	ENSG00000036257	CUL3	chr2	224470150	224585397	del
9407	ENSG00000187514	PTMA	chr2	231706895	231713541	del
9614	ENSG00000130294	KIF1A	chr2	240713761	240821036	del
10405	ENSG00000181555	SETD2	chr3	47016429	47163967	del
10504	ENSG00000067560	RHOA	chr3	49359145	49412998	del
11043	ENSG00000044524	EPHA3	chr3	89107621	89482134	amp
11808	ENSG00000051382	PIK3CB	chr3	138652699	138834938	del
11814	ENSG00000183770	FOXL2	chr3	138944224	138947137	del
12374	ENSG00000177565	TBL1XR1	chr3	177019340	177228000	amp
12402	ENSG00000121879	PIK3CA	chr3	179148114	179240093	amp
15434	ENSG00000083857	FAT1	chr4	186587794	186726722	amp
15551	ENSG00000164362	TERT	chr5	1253147	1295047	amp
16740	ENSG00000113318	MSH3	chr5	80654652	80876815	amp
17123	ENSG00000134982	APC	chr5	112707498	112846239	del
18512	ENSG00000164379	FOXQ1	chr6	1312098	1314758	amp
19285	ENSG00000234745	HLA-B	chr6	31269491	31357188	del
20143	ENSG00000156508	EEF1A1	chr6	73515750	73523797	del
20713	ENSG00000047936	ROS1	chr6	117288300	117425855	del
21001	ENSG00000118503	TNFAIP3	chr6	137867214	137883312	del
21164	ENSG00000131023	LATS1	chr6	149658153	149718105	del

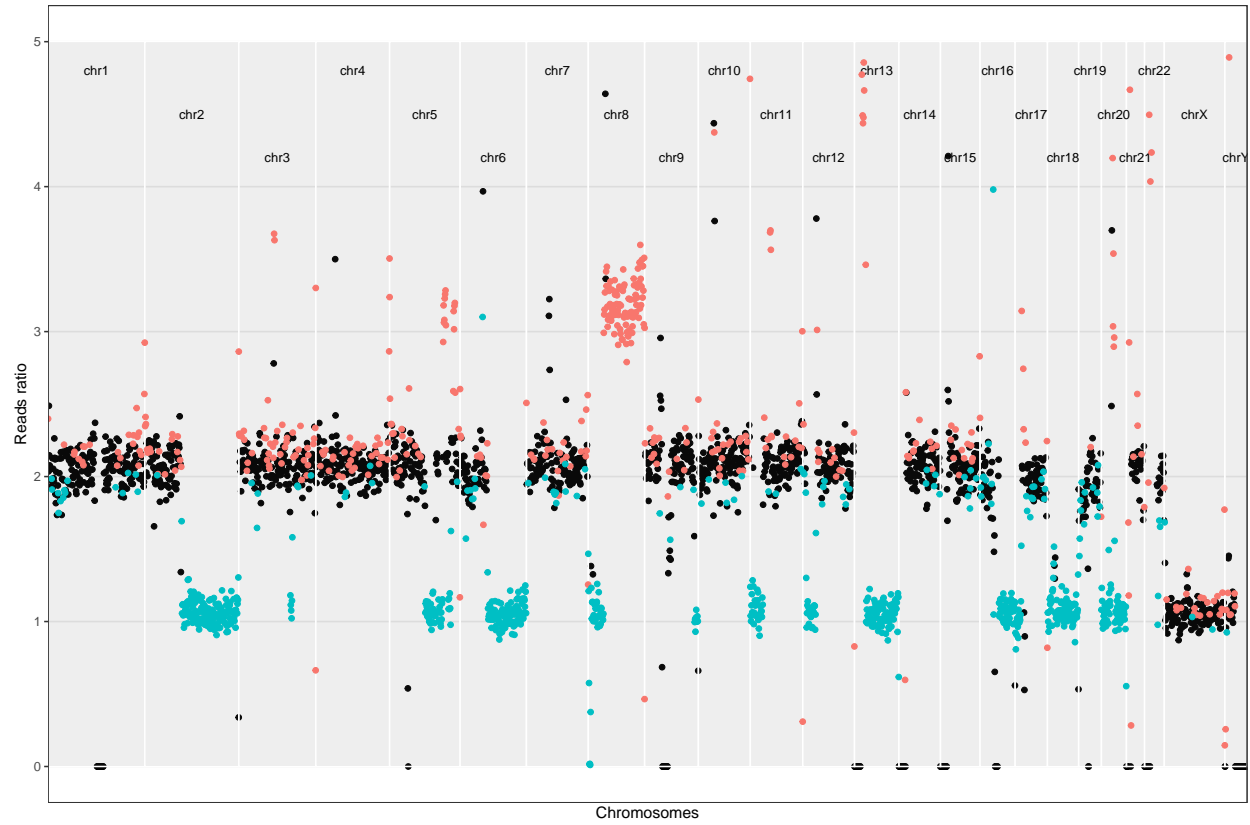
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21384	ENSG00000085511	MAP3K4	chr6	160991727	161117385	del
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22481	ENSG00000146648	EGFR	chr7	55019017	55211628	amp
23074	ENSG00000019991	HGF	chr7	81699010	81770438	amp
24217	ENSG00000197993	KEL	chr7	142941114	142962363	amp
24436	ENSG00000106615	RHEB	chr7	151466012	151520120	del
27499	ENSG00000179388	EGR3	chr8	22687659	22693480	del
27807	ENSG00000077782	FGFR1	chr8	38400215	38468834	del
28059	ENSG00000164736	SOX17	chr8	54457935	54460892	amp
28554	ENSG00000176571	CNBD1	chr8	86866442	87615219	amp
28965	ENSG00000164754	RAD21	chr8	116845935	116874866	amp
29133	ENSG00000136997	MYC	chr8	127735434	127742951	amp
29781	ENSG00000147889	CDKN2A	chr9	21967753	21995301	amp
30060	ENSG00000196092	PAX5	chr9	36833269	37034268	amp
31516	ENSG00000097007	ABL1	chr9	130713946	130887675	del
31558	ENSG00000165699	TSC1	chr9	132891348	132946874	del
31611	ENSG00000186350	RXRA	chr9	134317098	134440585	del
31616	ENSG00000130635	COL5A1	chr9	134641774	134844843	del
31674	ENSG00000148400	NOTCH1	chr9	136494433	136546048	del
31832	ENSG00000174775	HRAS	chr11	532242	537287	del
31870	ENSG00000184956	MUC6	chr11	1012823	1036718	del
32366	ENSG00000133818	RRAS2	chr11	14277922	14364506	del
32621	ENSG00000184937	WT1	chr11	32387775	32435885	del
35308	ENSG00000107485	GATA3	chr10	8045378	8075198	del
35859	ENSG00000165731	RET	chr10	43077069	43130351	amp
36150	ENSG00000150347	ARID5B	chr10	61901684	62096944	amp
37122	ENSG00000148737	TCF7L2	chr10	112950247	113167678	del
37379	ENSG00000170430	MGMT	chr10	129467190	129770983	amp
37950	ENSG00000111276	CDKN1B	chr12	12715058	12722369	del
37989	ENSG00000171681	ATF7IP	chr12	14365632	14502935	del
38141	ENSG00000133703	KRAS	chr12	25204789	25250936	del
38282	ENSG00000188375	H3F3C	chr12	31791185	31792298	del
38922	ENSG00000065361	ERBB3	chr12	56076799	56103505	del
39959	ENSG00000179295	PTPN11	chr12	112418351	112509913	del
40154	ENSG00000135100	HNF1A	chr12	120978543	121002512	del
40502	ENSG00000177084	POLE	chr12	132623753	132687365	amp
40605	ENSG00000121741	ZMYM2	chr13	19958670	20091829	amp
40632	ENSG00000150457	LATS2	chr13	20973036	21061586	amp
40795	ENSG00000122025	FLT3	chr13	28003274	28100592	amp
40868	ENSG00000139618	BRCA2	chr13	32315086	32400266	del
40876	ENSG00000083642	PDS5B	chr13	32586452	32778019	del
41134	ENSG00000139687	RB1	chr13	48303726	48599436	del
41145	ENSG00000152207	CYSLTR2	chr13	48653711	48711226	del
41388	ENSG00000276644	DACH1	chr13	71437966	71867204	del
41406	ENSG00000102554	KLF5	chr13	73054976	73077541	del
42295	ENSG00000129474	AJUBA	chr14	22971177	22982551	amp
42601	ENSG00000129514	FOXA1	chr14	37589552	37596059	amp
46467	ENSG00000103126	AXIN1	chr16	287440	352723	amp
46622	ENSG00000103197	TSC2	chr16	2047967	2089491	del
46775	ENSG00000005339	CREBBP	chr16	3725054	3880726	del
47953	ENSG00000166164	BRD7	chr16	50313487	50368934	del

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47966	ENSG00000083799	CYLD	chr16	50742050	50801935	del
48110	ENSG00000102900	NUP93	chr16	56730118	56850286	del
48288	ENSG00000067955	CBFB	chr16	67028984	67101058	del
48327	ENSG00000102974	CTCF	chr16	67562467	67639177	del
48396	ENSG00000039068	CDH1	chr16	68737292	68835541	del
48539	ENSG00000140836	ZFHX3	chr16	72782885	73891871	del
49313	ENSG00000132522	GPS2	chr17	7311324	7315564	del
49360	ENSG00000141510	TP53	chr17	7661779	7687550	del
49370	ENSG00000170004	CHD3	chr17	7884806	7912760	del
49491	ENSG00000065559	MAP2K4	chr17	12020829	12143830	del
49580	ENSG00000141027	NCOR1	chr17	16029157	16218185	del
50158	ENSG00000196712	NF1	chr17	31094927	31382116	del
50479	ENSG00000167258	CDK12	chr17	39461486	39564907	del
50489	ENSG00000141736	ERBB2	chr17	39687914	39730426	del
50883	ENSG00000120071	KANSL1	chr17	46029916	46225389	del
51040	ENSG00000121067	SPOP	chr17	49598884	49678163	del
51550	ENSG00000108946	PRKAR1A	chr17	68511780	68551319	del
52042	ENSG00000141579	ZNF750	chr17	82829434	82840022	amp
52137	ENSG00000177426	TGIF1	chr18	3411608	3459978	del
52741	ENSG00000152217	SETBP1	chr18	44680173	45068510	del
52799	ENSG00000175387	SMAD2	chr18	47808957	47931146	del
52876	ENSG00000141646	SMAD4	chr18	51028394	51085045	del
53051	ENSG00000171791	BCL2	chr18	63123346	63320128	del
53512	ENSG00000101333	PLCB4	chr20	9068763	9481242	del
53638	ENSG00000125846	ZNF133	chr20	18288283	18316996	del
53723	ENSG00000125798	FOXA2	chr20	22581005	22585455	del
53921	ENSG00000171456	ASXL1	chr20	32358330	32439319	amp
54089	ENSG00000197122	SRC	chr20	37344685	37406050	del
54163	ENSG00000124181	PLCG1	chr20	41136960	41196801	del
54499	ENSG00000087586	AURKA	chr20	56369389	56392337	del
54566	ENSG00000087460	GNAS	chr20	58839718	58911192	del
54949	ENSG00000088256	GNA11	chr19	3094362	3123999	del
54995	ENSG00000167658	EEF2	chr19	3976056	3985463	del
55117	ENSG00000125726	CD70	chr19	6583183	6604103	del
56943	ENSG00000105464	GRIN2D	chr19	48394875	48444931	del
58538	ENSG00000099949	LZTR1	chr22	20982269	20999032	amp
58601	ENSG00000100030	MAPK1	chr22	21754500	21867680	amp
58779	ENSG00000099956	SMARCB1	chr22	23786931	23838008	amp
58935	ENSG00000183765	CHEK2	chr22	28687743	28742422	amp
58970	ENSG00000186575	NF2	chr22	29603556	29698598	amp
249381	ENSG00000198947	DMD	chrX	31097677	33339441	amp
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263741	ENSG00000101972	STAG2	chrX	123960212	124422664	del

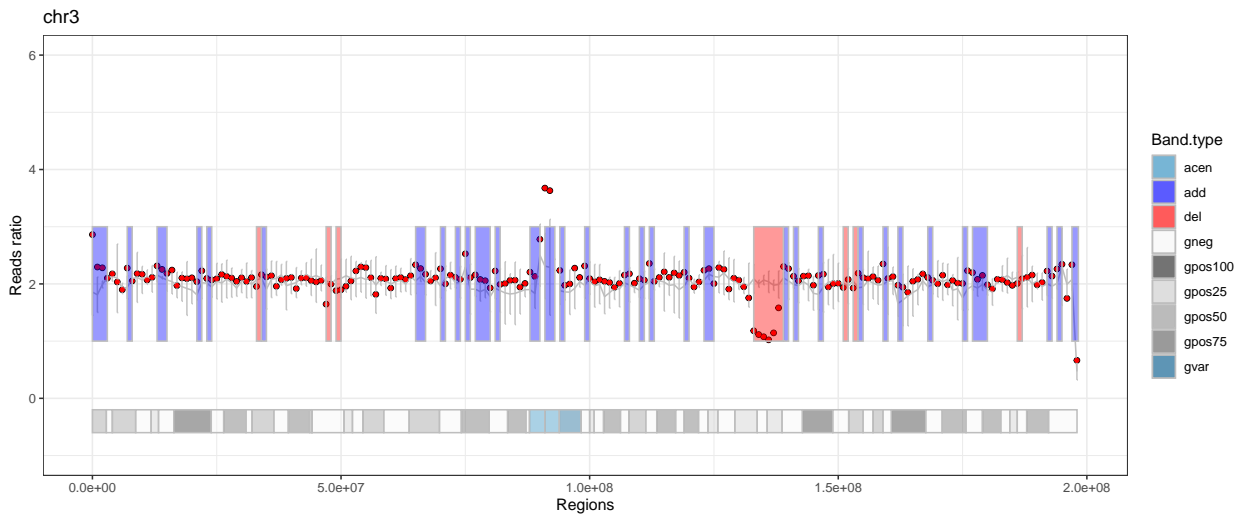
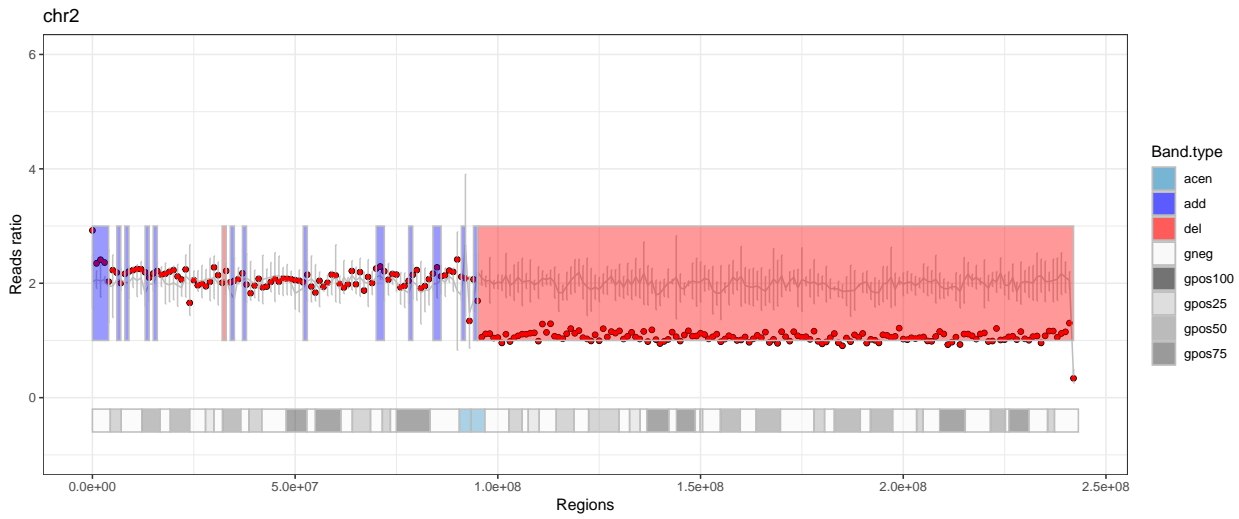
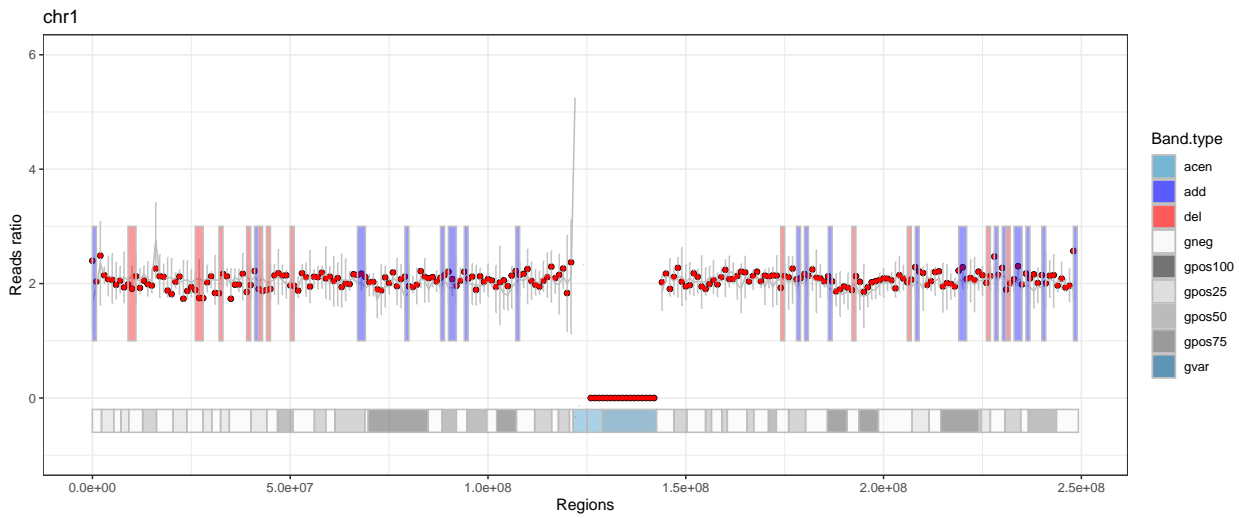
Bailey et al. (2018): Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell 173 (2), 371-385.e18. DOI: 10.1016/j.cell.2018.02.060.;

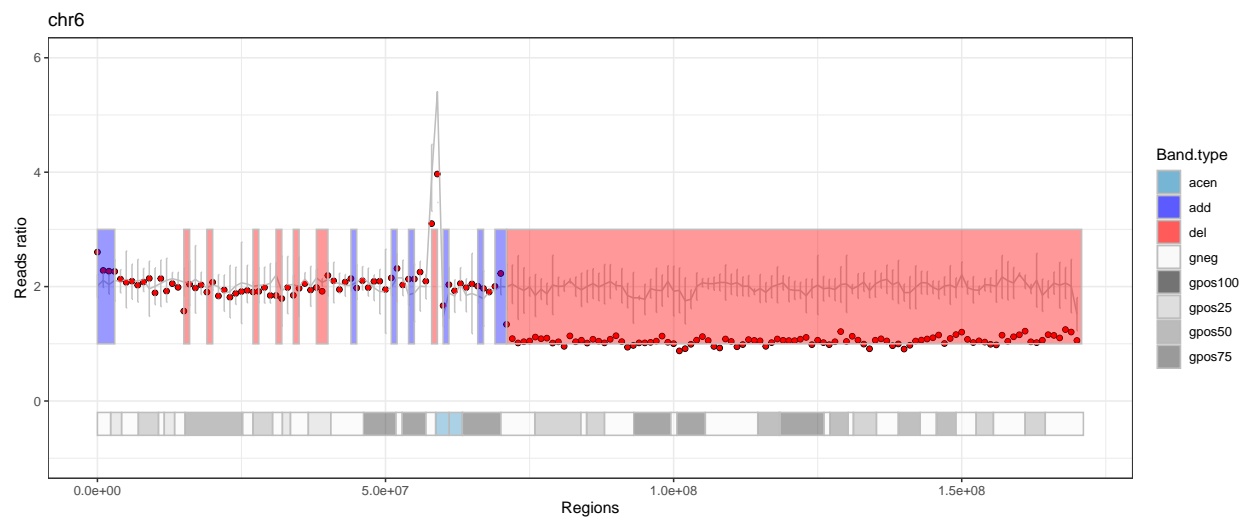
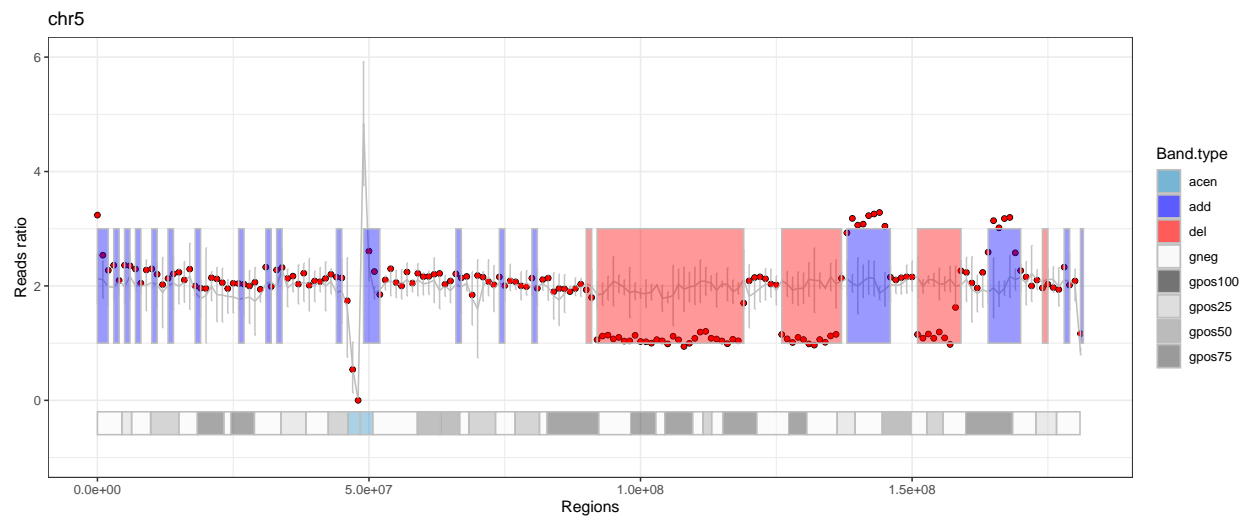
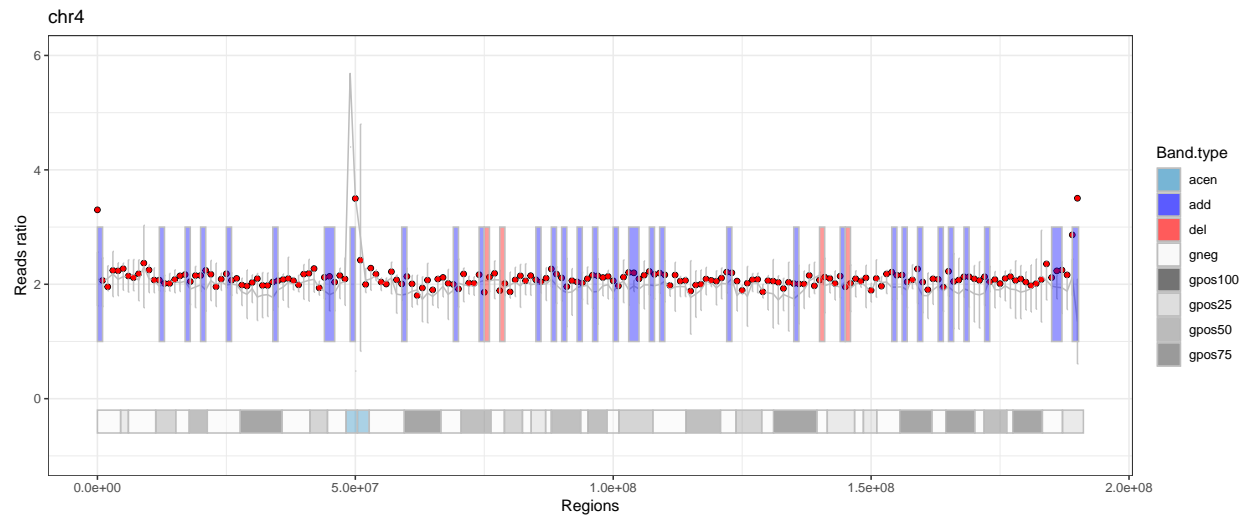
Papaemmanuil et al. (2016): Genomic Classification and Prognosis in Acute Myeloid Leukemia. The New England journal of medicine 374 (23), S. 2209–2221. DOI:10.1056/NEJMoa1516192.

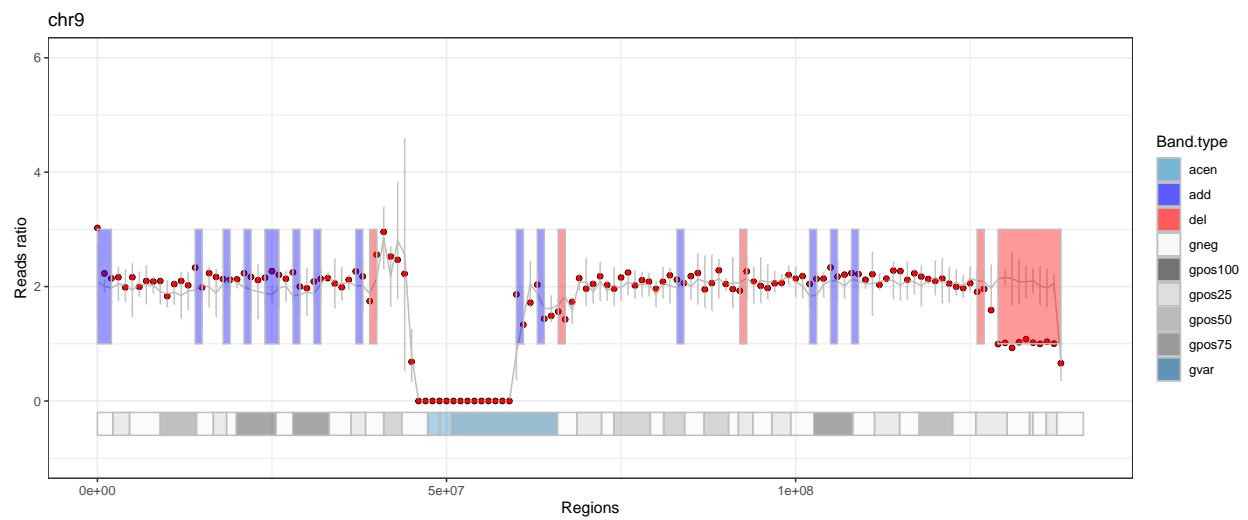
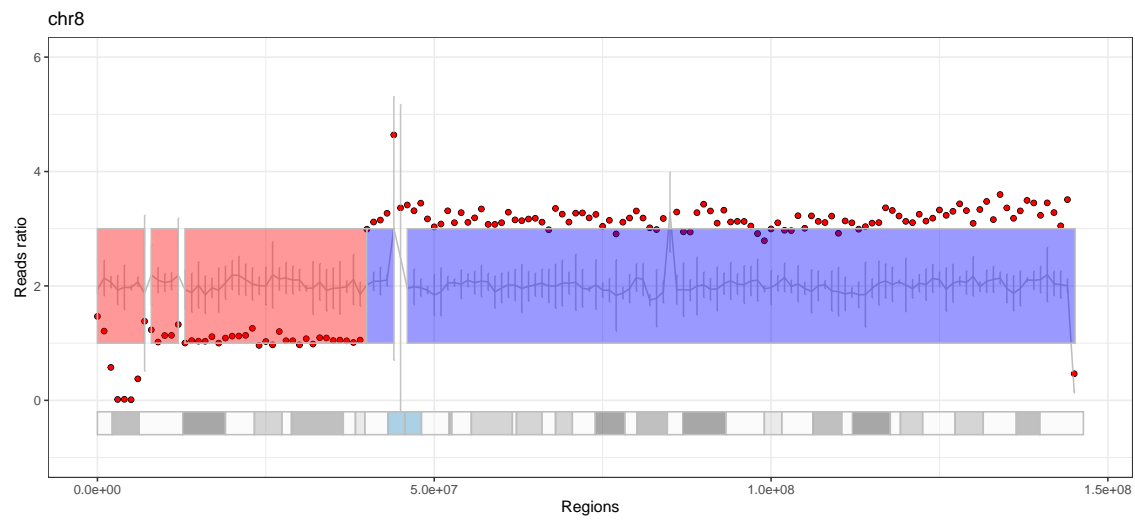
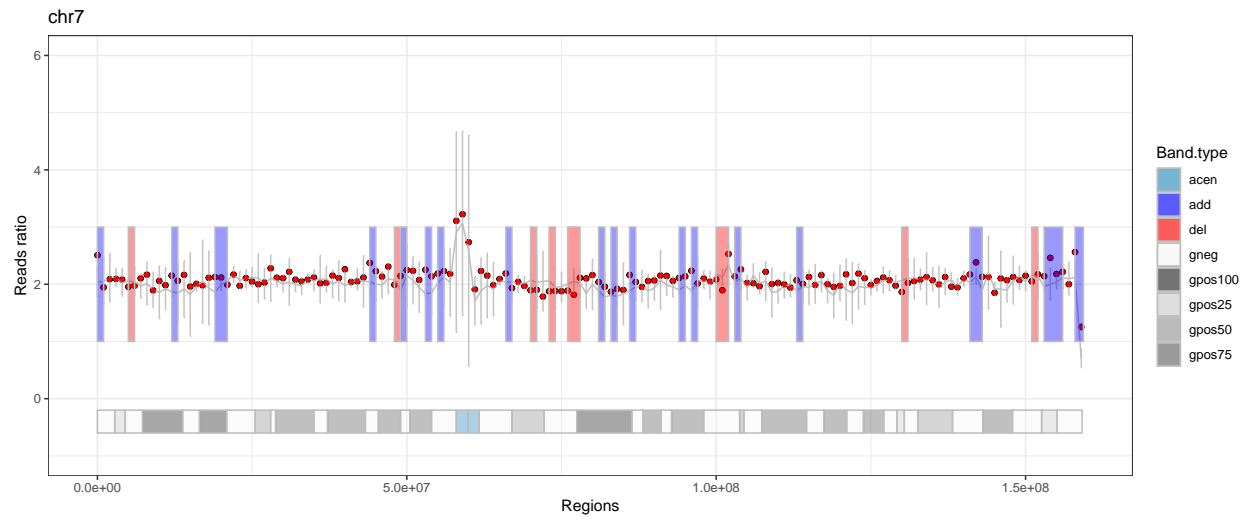
Karyotype overview:

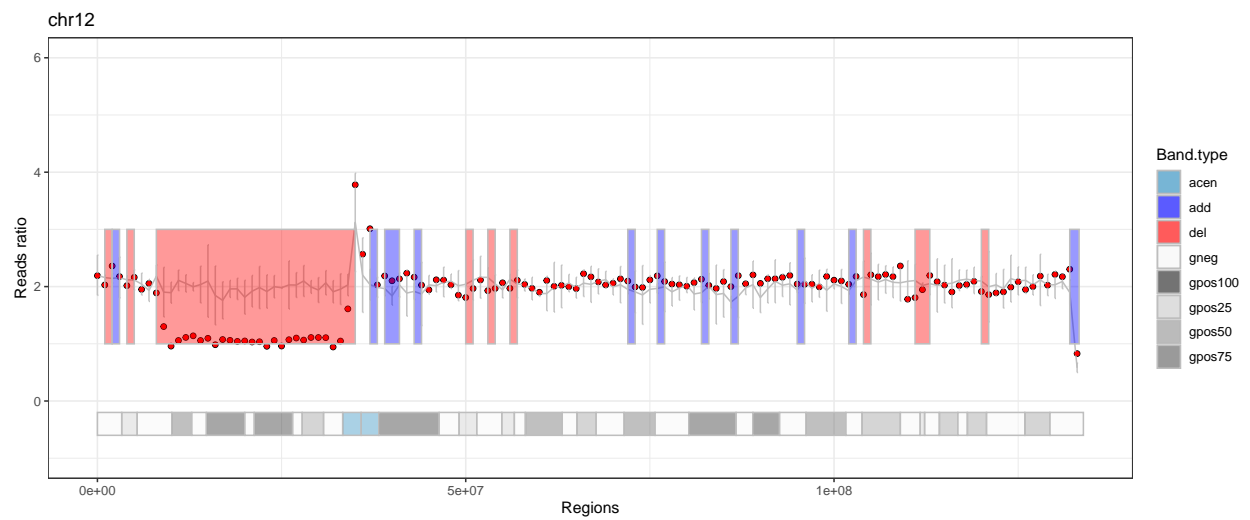
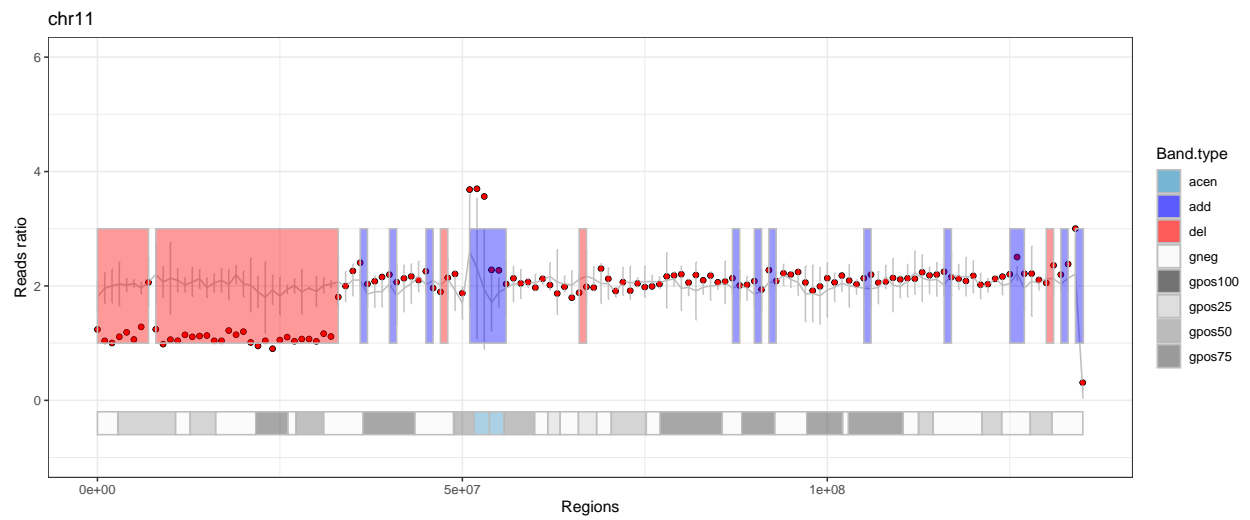
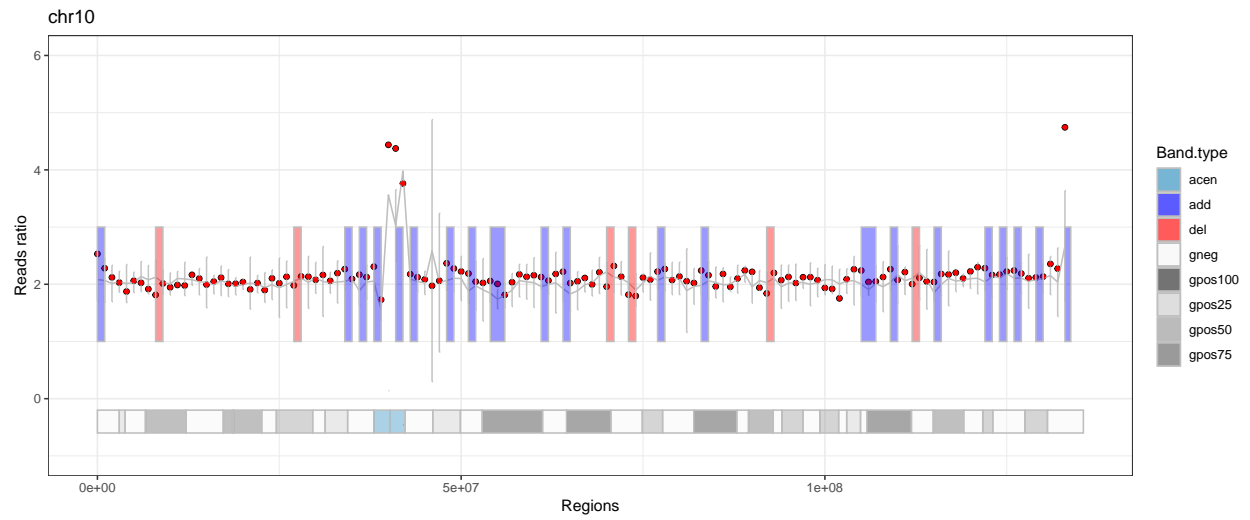


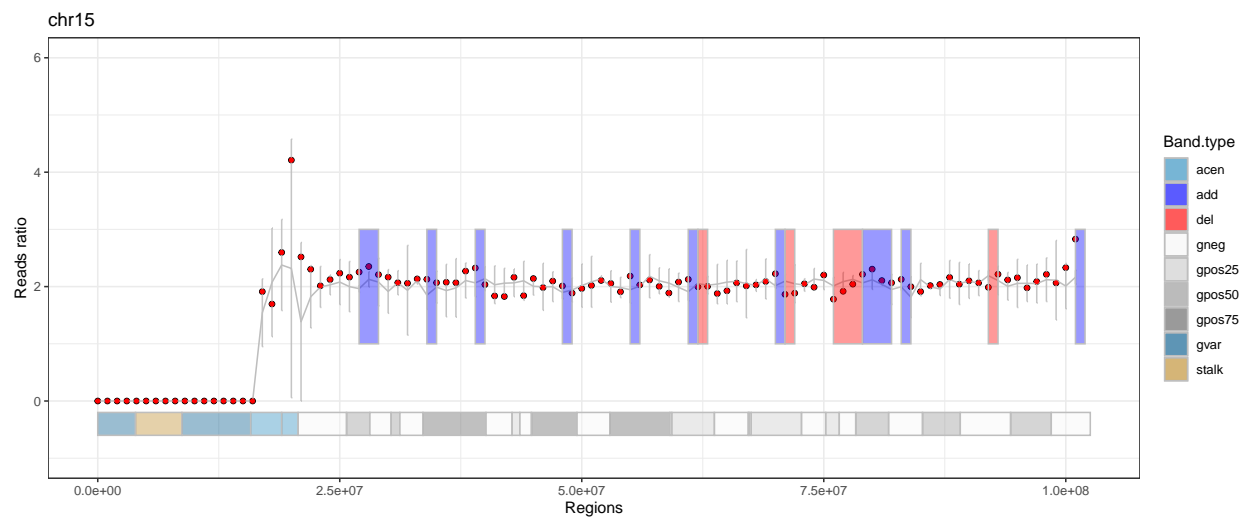
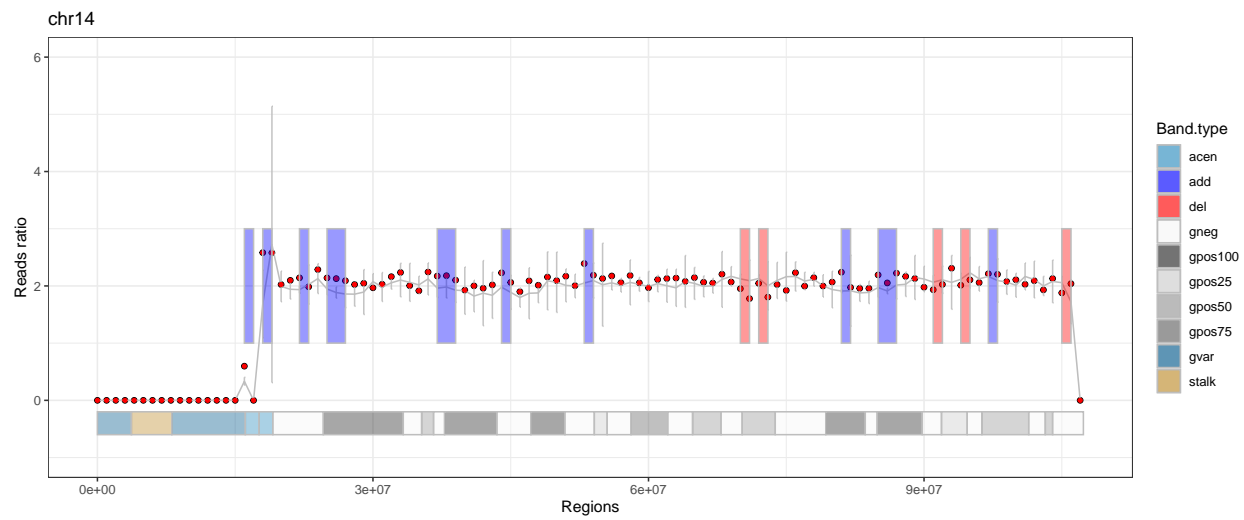
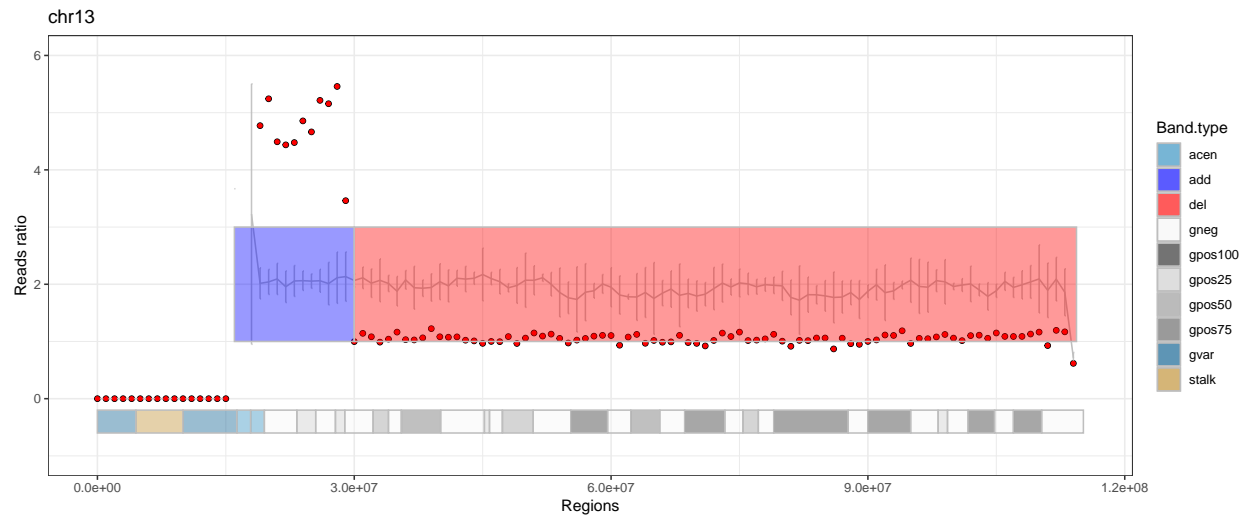
Chromosome Plots:

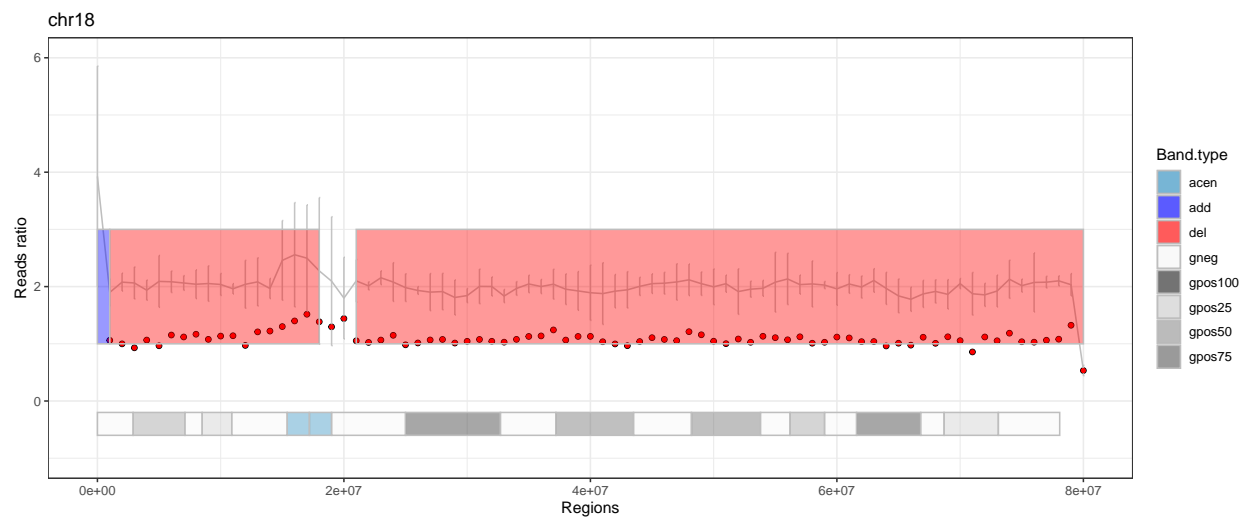
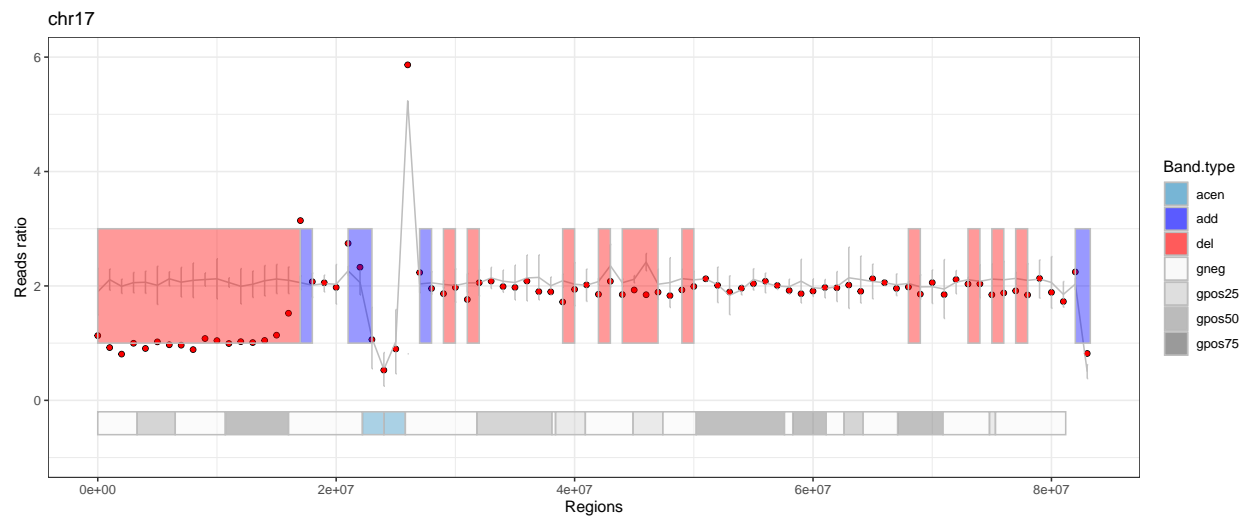
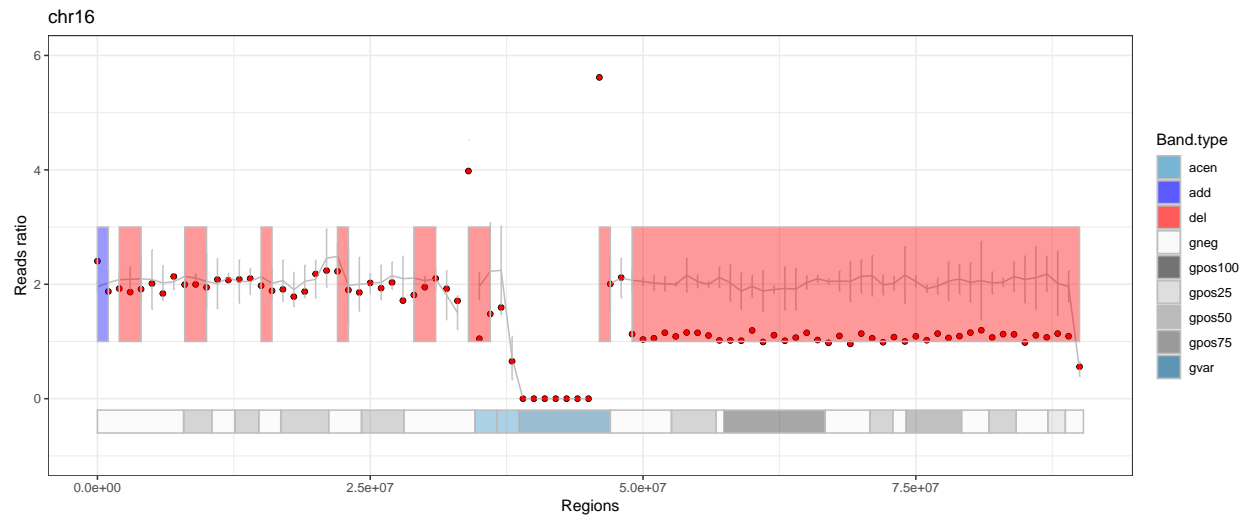


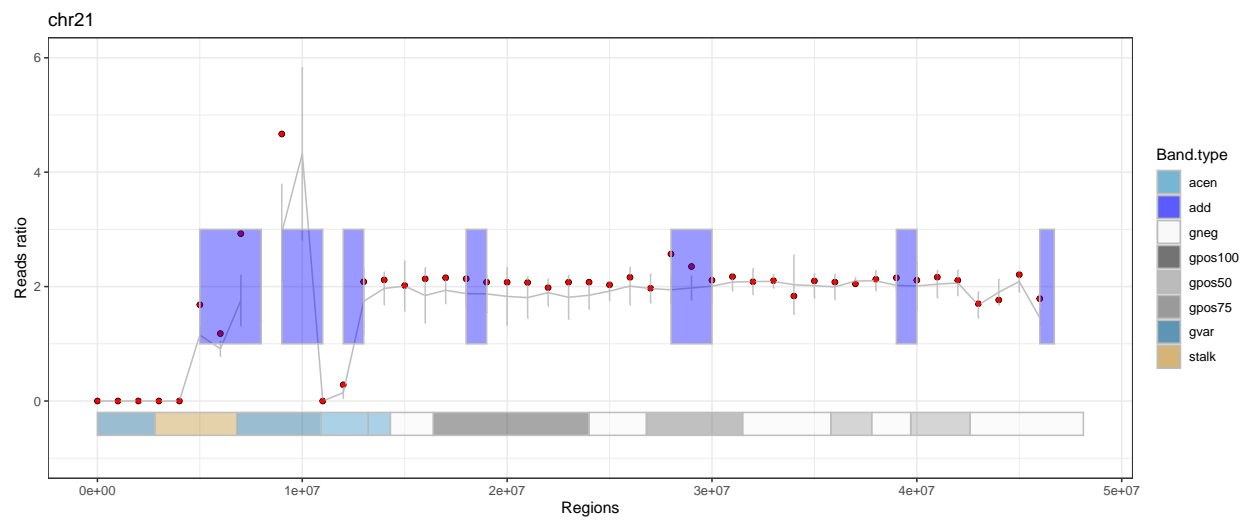
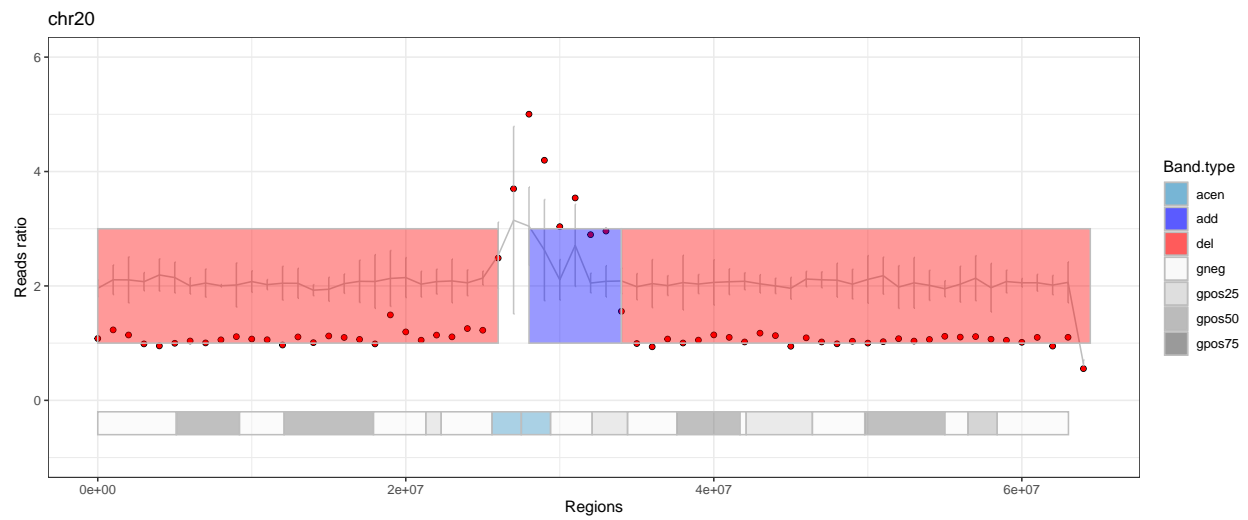
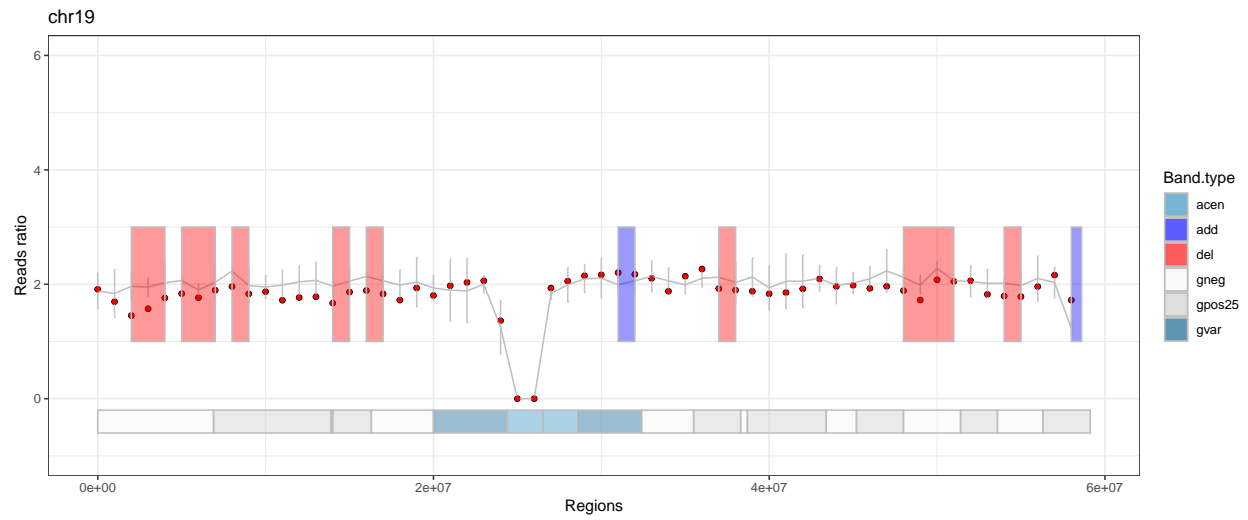


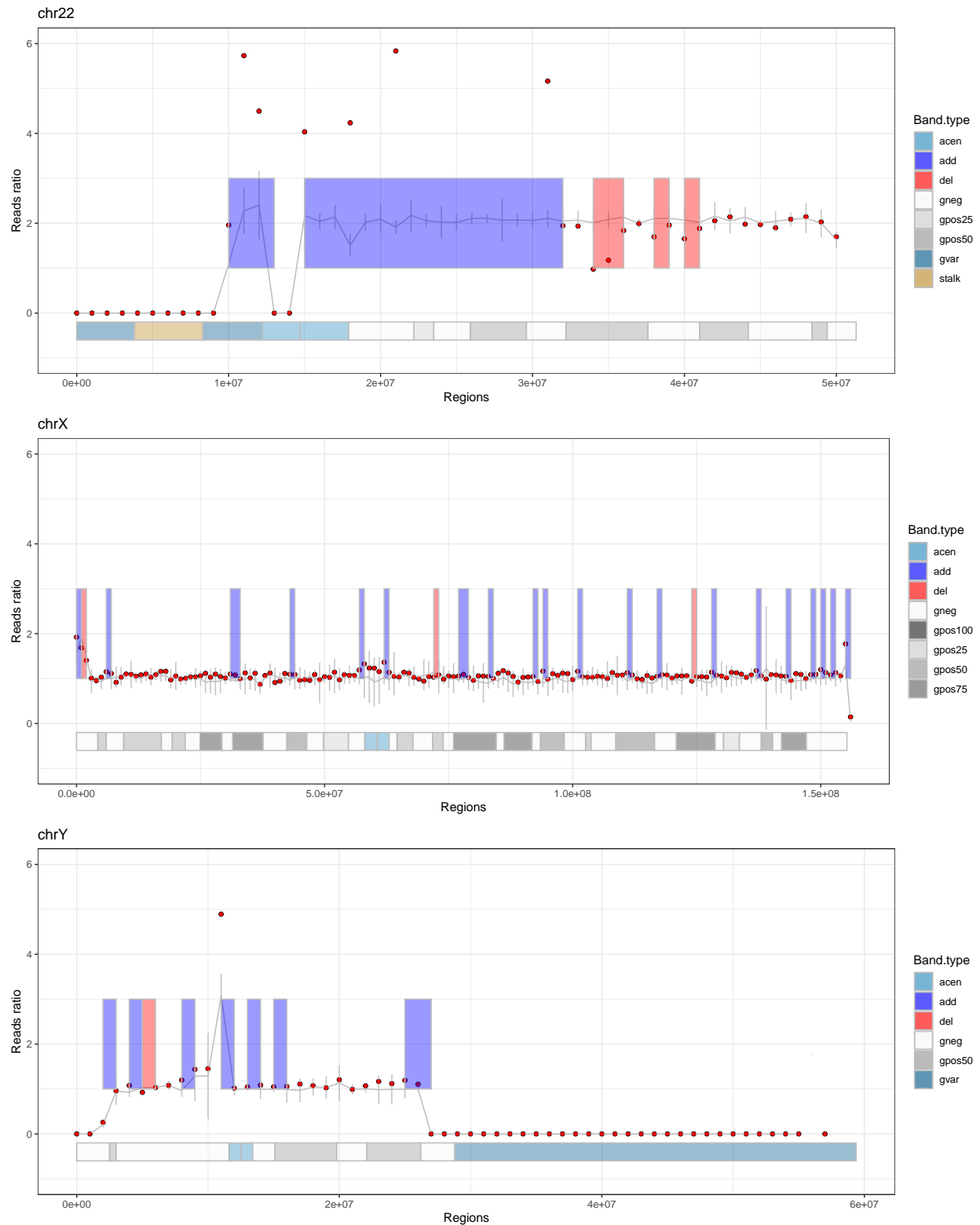












The legend contains two characteristics:

1. Giemsa stain results. Recognized stain values: gneg, gpos50, gpos75, gpos25, gpos100, acen,

- gvar, stalk.
2. The abnormal regions (add or del).