

Report CORIANDR: ChrOmosomal abeRration Identifier AND Reporter in R

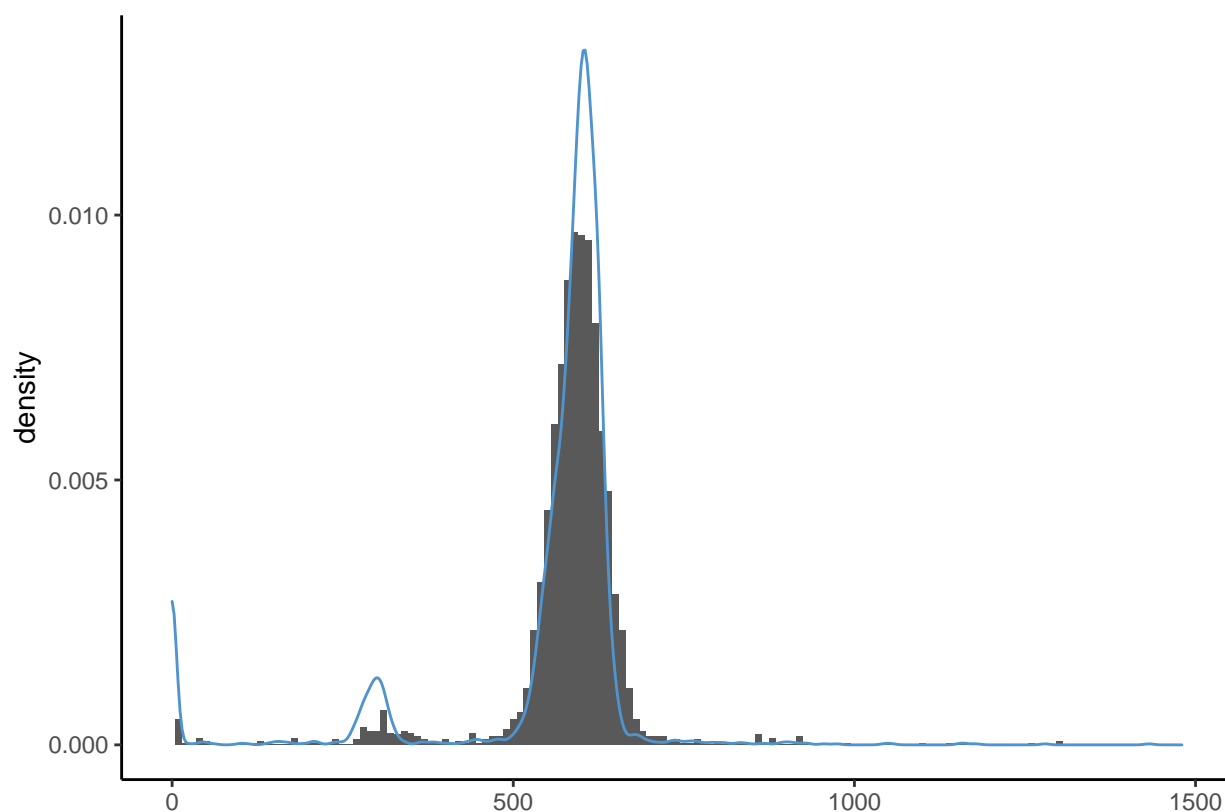
03 März, 2021

Sample ID

98217.new.report_2

row read pairs	897816
average read length	149.281
unique mapped pairs	874972
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:

del(1)(p36.31p36.31)
del(1)(p36.12p36.11)
del(1)(p35.1p35.1)
del(1)(p34.2p34.2)
del(1)(p31.3p31.3)
del(1)(q24.1q24.1)
add(1)(q31.1q31.2)
del(1)(q42.12q42.2)
del(2)(p25.1p25.1)
del(2)(p22.2p22.2)
add(2)(p16.3p16.3)
add(2)(p16.1p16.1)
add(2)(p11.2p11.2)
del(2)(q21.2q21.2)
add(2)(q32.1q32.1)
del(2)(q32.2q32.2)
add(2)(q34q34)
del(2)(q36.1q36.1)
add(3)(p26.3p26.3)
del(3)(p26.2p26.2)
del(3)(p25.2p25.1)
add(3)(p24.3p24.3)
del(3)(p23p23)
add(3)(p22.3p22.3)
del(3)(p21.32p21.32)
del(3)(p14.3p14.3)
del(3)(p14.1p14.1)
add(3)(p12.2p12.2)
add(3)(p11.2p11.2)
add(3)(q11.1q11.2)
add(3)(q13.2q13.2)
del(3)(q21.1q21.1)
add(3)(q24q24)
add(3)(q25.31q25.31)
del(3)(q25.32q25.32)

add(3)(q26.1q26.1)
 del(3)(q27.2q27.2)
 del(4)(p16.1p16.1)
 add(4)(p12p12)
 add(4)(q13.1q13.1)
 del(4)(q21.3q21.3)
 add(4)(q22.2q22.2)
 add(4)(q28.2q28.2)
 del(4)(q31.1q31.1)
 add(4)(q34.3q34.3)
 del(5)(p13.1p13.1)
 del(5)(q14.1q14.1)
 add(5)(q14.3q21.2)
 add(5)(q22.1q22.1)
 del(5)(q33.2q33.3)
 del(5)(q35.1q35.2)
 del(6)(p21.33p21.33)
 del(6)(p21.1p21.1)
 del(6)(p11.2p11.2)
 add(6)(q13q14.1)
 add(6)(q16.1q16.1)
 add(6)(q22.1q22.1)
 add(6)(q22.33q22.33)
 del(6)(q23.2q23.3)
 add(6)(q25.2q25.2)
 del(6)(q25.3q25.3)
 add(7)(p12.2p12.1)
 del(7)(q11.21q11.21)
 del(7)(q21.11q36.3)
 del(8)(p21.3p21.3)
 del(8)(p21.1p12)
 add(8)(q12.3q12.3)
 add(8)(q21.12q21.13)
 del(9)(p24.1p24.1)
 add(9)(p21.2p21.2)
 del(9)(q21.11q21.11)

$\text{del}(9)(q_{33.1}q_{33.1})$
 $\text{del}(9)(q_{33.3}q_{33.3})$
 $\text{add}(10)(p_{11.1}p_{11.1})$
 $\text{add}(10)(q_{21.1}q_{21.1})$
 $\text{del}(10)(q_{22.1}q_{22.1})$
 $\text{del}(10)(q_{26.11}q_{26.11})$
 $\text{del}(10)(q_{26.13}q_{26.13})$
 $\text{del}(11)(p_{15.2}p_{15.1})$
 $\text{add}(11)(p_{14.2}p_{14.1})$
 $\text{del}(11)(p_{11.2}p_{11.12})$
 $\text{del}(11)(q_{13.4}q_{13.4})$
 $\text{del}(11)(q_{14.2}q_{14.2})$
 $\text{del}(11)(q_{23.2}q_{23.2})$
 $\text{add}(12)(p_{12.3}p_{12.3})$
 $\text{add}(12)(p_{12.1}p_{12.1})$
 $\text{del}(12)(q_{13.12}q_{13.12})$
 $\text{del}(12)(q_{13.2}q_{13.3})$
 $\text{add}(12)(q_{21.32}q_{21.32})$
 $\text{del}(12)(q_{23.3}q_{23.3})$
 $\text{del}(12)(q_{24.12}q_{24.12})$
 $\text{del}(12)(q_{24.23}q_{24.23})$
 $\text{add}(13)(q_{11}q_{11})$
 $\text{del}(13)(q_{12.12}q_{12.12})$
 $\text{del}(13)(q_{14.11}q_{14.11})$
 $\text{add}(13)(q_{21.33}q_{21.33})$
 $\text{add}(13)(q_{22.3}q_{22.3})$
 $\text{add}(13)(q_{33.2}q_{33.2})$
 $\text{add}(14)(q_{12}q_{12})$
 $\text{add}(14)(q_{21.2}q_{21.2})$
 $\text{del}(14)(q_{23.1}q_{23.1})$
 $\text{del}(14)(q_{24.2}q_{24.2})$
 $\text{add}(14)(q_{31.2}q_{31.2})$
 $\text{del}(14)(q_{32.11}q_{32.11})$
 $\text{add}(15)(q_{21.1}q_{21.1})$
 $\text{del}(15)(q_{21.2}q_{21.3})$
 $\text{add}(15)(q_{22.2}q_{22.2})$

add(15)(q25.2q25.2)
 del(15)(q26.1q26.1)
 del(16)(p13.11p12.3)
 del(16)(p11.1p11.1)
 del(16)(q12.2q12.2)
 add(16)(q13q13)
 del(16)(q22.1q22.1)
 add(16)(q22.3q22.3)
 del(17)(p13.3p13.3)
 add(17)(p11.1p11.1)
 del(17)(q12q12)
 del(17)(q21.32q21.32)
 del(17)(q22q22)
 del(18)(q11.2q11.2)
 add(18)(q12.2q12.2)
 add(18)(q21.2q21.2)
 add(18)(q22.2q22.3)
 del(19)(p13.12p13.12)
 del(19)(q13.11q13.11)
 del(19)(q13.33q13.41)
 add(20)(p12.1p12.1)
 del(20)(p11.21p11.21)
 del(20)(q13.13q13.13)
 add(21)(q21.3q21.3)
 add(21)(q22.13q22.13)
 add(22)(q11.21q11.21)
 del(22)(q12.1q12.2)
 del(22)(q13.1q13.1)
 add(22)(q13.33q13.33)
 add(X)(p22.33q28)
 del(Y)(p11.31q12)

Genes affected by CNVs*:

id	sym	bol chr	sta	rt end	abe	rration
882	ENSG00000117713	ARID1A	chr1	26693236	26782104	del
1201	ENSG00000163874	ZC3H12A	chr1	37474580	37484377	del

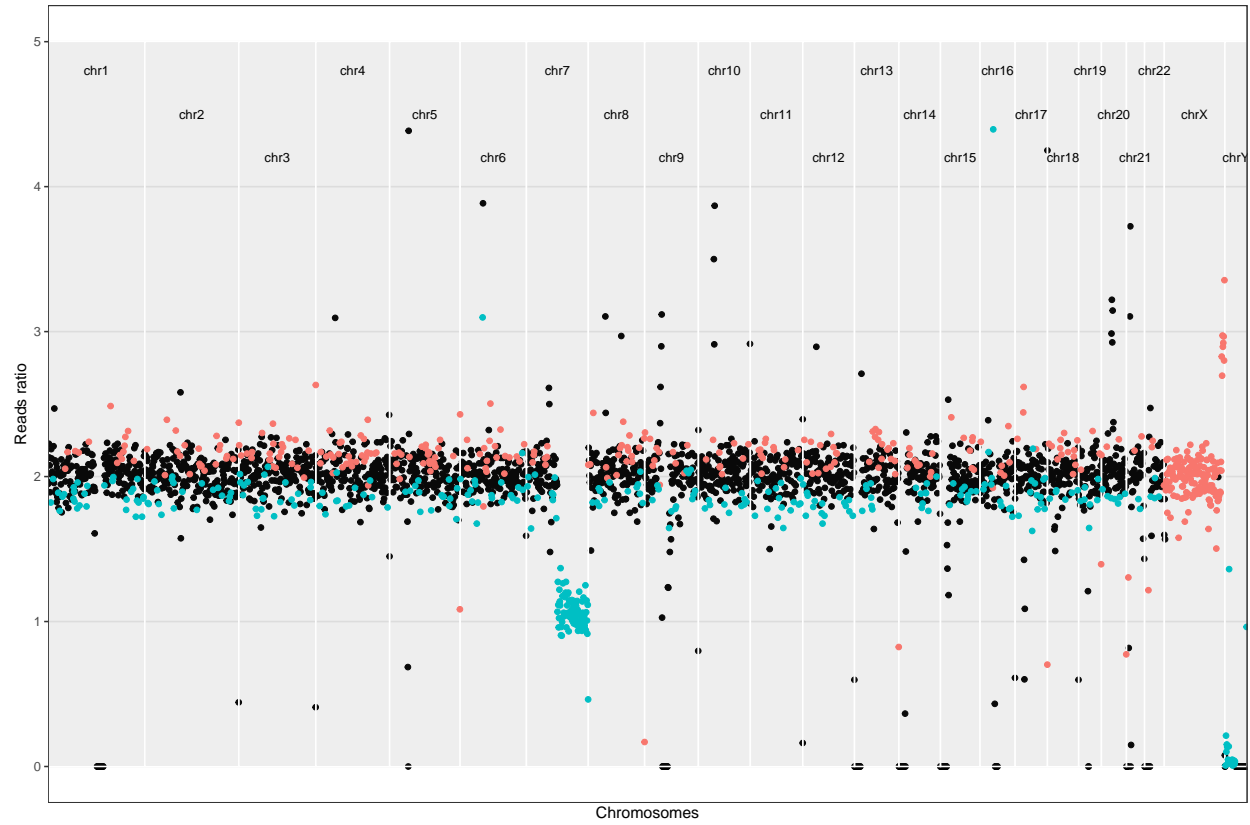
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1604	ENSG00000123080	CDKN2C	chr1	50960745	50974634	amp
2967	ENSG00000265972	TXNIP	chr1	145992435	145996600	del
4392	ENSG00000163435	ELF3	chr1	202007945	202017183	del
4895	ENSG00000163041	H3F3A	chr1	226061851	226072007	del
5025	ENSG00000069248	NUP133	chr1	229440259	229508341	del
5860	ENSG00000119772	DNMT3A	chr2	25227855	25342590	del
5866	ENSG00000143970	ASXL2	chr2	25733753	25878516	del
6183	ENSG00000152518	ZFP36L2	chr2	43222402	43226606	del
6626	ENSG00000169564	PCBP1	chr2	70087454	70089203	del
8082	ENSG00000121989	ACVR2A	chr2	147844517	147930826	del
8662	ENSG00000064933	PMS1	chr2	189784085	189877629	del
9038	ENSG00000178568	ERBB4	chr2	211375717	212538841	amp
10405	ENSG00000181555	SETD2	chr3	47016429	47163967	del
11043	ENSG00000044524	EPHA3	chr3	89107621	89482134	amp
11346	ENSG00000181722	ZBTB20	chr3	114314501	115147271	amp
13641	ENSG00000134853	PDGFRA	chr4	54229280	54298245	del
13646	ENSG00000157404	KIT	chr4	54657918	54740715	del
14974	ENSG00000109670	FBXW7	chr4	152320544	152536092	amp
16825	ENSG00000145715	RASA1	chr5	87267888	87391931	amp
18512	ENSG00000164379	FOXQ1	chr6	1312098	1314758	del
19285	ENSG00000234745	HLA-B	chr6	31269491	31357188	del
21384	ENSG00000085511	MAP3K4	chr6	160991727	161117385	del
21626	ENSG00000198286	CARD11	chr7	2906141	3043945	del
21681	ENSG00000122512	PMS2	chr7	5970925	6009106	del
23074	ENSG00000019991	HGF	chr7	81699010	81770438	del
23552	ENSG00000105851	PIK3CG	chr7	106865278	106908980	del
23659	ENSG00000105976	MET	chr7	116672196	116798386	del
23865	ENSG00000128602	SMO	chr7	129188633	129213545	del
24082	ENSG00000157764	BRAF	chr7	140719327	140924928	del
24217	ENSG00000197993	KEL	chr7	142941114	142962363	del
24328	ENSG00000055130	CUL1	chr7	148697914	148801110	del
24329	ENSG00000106462	EZH2	chr7	148807383	148884321	del
24436	ENSG00000106615	RHEB	chr7	151466012	151520120	del
24449	ENSG00000055609	KMT2C	chr7	152134922	152436005	del
27499	ENSG00000179388	EGR3	chr8	22687659	22693480	del
28554	ENSG00000176571	CNBD1	chr8	86866442	87615219	amp
29781	ENSG00000147889	CDKN2A	chr9	21967753	21995301	amp
31516	ENSG00000097007	ABL1	chr9	130713946	130887675	del
32621	ENSG00000184937	WT1	chr11	32387775	32435885	amp
36150	ENSG00000150347	ARID5B	chr10	61901684	62096944	amp
37122	ENSG00000148737	TCF7L2	chr10	112950247	113167678	del
37379	ENSG00000170430	MGMT	chr10	129467190	129770983	amp
38141	ENSG00000133703	KRAS	chr12	25204789	25250936	amp
38282	ENSG00000188375	H3F3C	chr12	31791185	31792298	del
38444	ENSG00000189079	ARID2	chr12	45729706	45908040	amp
38568	ENSG00000167548	KMT2D	chr12	49018975	49059774	del
38922	ENSG00000065361	ERBB3	chr12	56076799	56103505	del
40154	ENSG00000135100	HNF1A	chr12	120978543	121002512	del
40605	ENSG00000121741	ZMYM2	chr13	19958670	20091829	del
40632	ENSG00000150457	LATS2	chr13	20973036	21061586	del
41388	ENSG00000276644	DACH1	chr13	71437966	71867204	amp
42295	ENSG00000129474	AJUBA	chr14	22971177	22982551	amp

id	sym	bol chr	sta	rt end	abe	rration
45273	ENSG00000140262	TCF12	chr15	56918623	57299281	del
46622	ENSG00000103197	TSC2	chr16	2047967	2089491	amp
48110	ENSG00000102900	NUP93	chr16	56730118	56850286	amp
48288	ENSG00000067955	CBFB	chr16	67028984	67101058	del
48327	ENSG00000102974	CTCF	chr16	67562467	67639177	del
48539	ENSG00000140836	ZFHX3	chr16	72782885	73891871	amp
50479	ENSG00000167258	CDK12	chr17	39461486	39564907	del
50489	ENSG00000141736	ERBB2	chr17	39687914	39730426	del
50883	ENSG00000120071	KANSL1	chr17	46029916	46225389	del
51230	ENSG00000108375	RNF43	chr17	58352500	58417595	del
51776	ENSG00000161547	SRSF2	chr17	76734115	76737374	del
53051	ENSG00000171791	BCL2	chr18	63123346	63320128	del
55117	ENSG00000125726	CD70	chr19	6583183	6604103	amp
56102	ENSG00000105173	CCNE1	chr19	29811991	29824312	del
56943	ENSG00000105464	GRIN2D	chr19	48394875	48444931	del
58601	ENSG00000100030	MAPK1	chr22	21754500	21867680	amp
59615	ENSG00000196576	PLXNB2	chr22	50274979	50307646	amp
60103	ENSG00000156304	SCAF4	chr21	31671033	31732075	del
247031	ENSG00000205542	TMSB4X	chrX	12975110	12977227	amp
248091	ENSG00000173674	EIF1AX	chrX	20124525	20141838	amp
248131	ENSG00000177189	RPS6KA3	chrX	20149911	20267519	amp
249381	ENSG00000198947	DMD	chrX	31097677	33339441	amp
250291	ENSG00000183337	BCOR	chrX	40049815	40177329	amp
250501	ENSG00000124486	USP9X	chrX	41085445	41236579	amp
250551	ENSG00000215301	DDX3X	chrX	41333284	41364472	amp
251001	ENSG00000147050	KDM6A	chrX	44873177	45112602	amp
251431	ENSG00000182872	RBM10	chrX	47145221	47186813	amp
251601	ENSG00000078061	ARAF	chrX	47561100	47571920	amp
253891	ENSG00000126012	KDM5C	chrX	53191321	53225422	amp
253981	ENSG00000072501	SMC1A	chrX	53374149	53422728	amp
254051	ENSG00000086758	HUWE1	chrX	53532096	53686728	amp
254971	ENSG00000184675	AMER1	chrX	64185117	64205708	amp
255391	ENSG00000169083	AR	chrX	67544021	67730619	amp
256071	ENSG00000184634	MED12	chrX	71118556	71142454	amp
256111	ENSG00000147130	ZMYM3	chrX	71239624	71255146	amp
256161	ENSG00000147133	TAF1	chrX	71366239	71532374	amp
257591	ENSG00000085224	ATRX	chrX	77504878	77786233	amp
259151	ENSG00000147202	DIAPH2	chrX	96684663	97604997	amp
262571	ENSG00000174460	ZCCHC12	chrX	118823824	118826968	amp
263741	ENSG00000101972	STAG2	chrX	123960212	124422664	amp
264151	ENSG00000102038	SMARCA1	chrX	129446501	129523500	amp
265031	ENSG00000156531	PHF6	chrX	134373253	134428791	amp
268991	ENSG00000196924	FLNA	chrX	154348524	154374638	amp

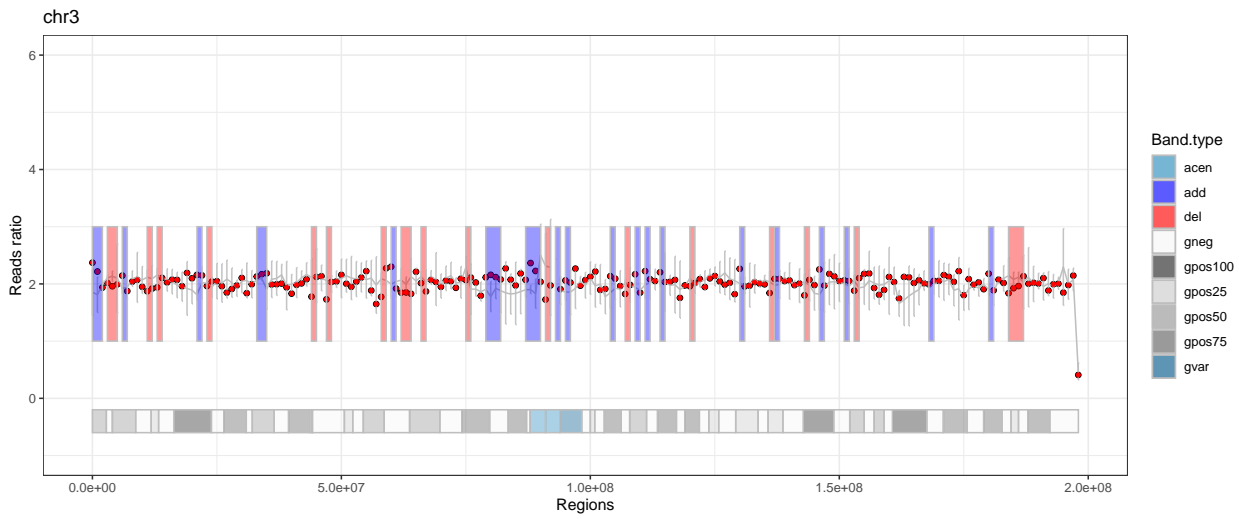
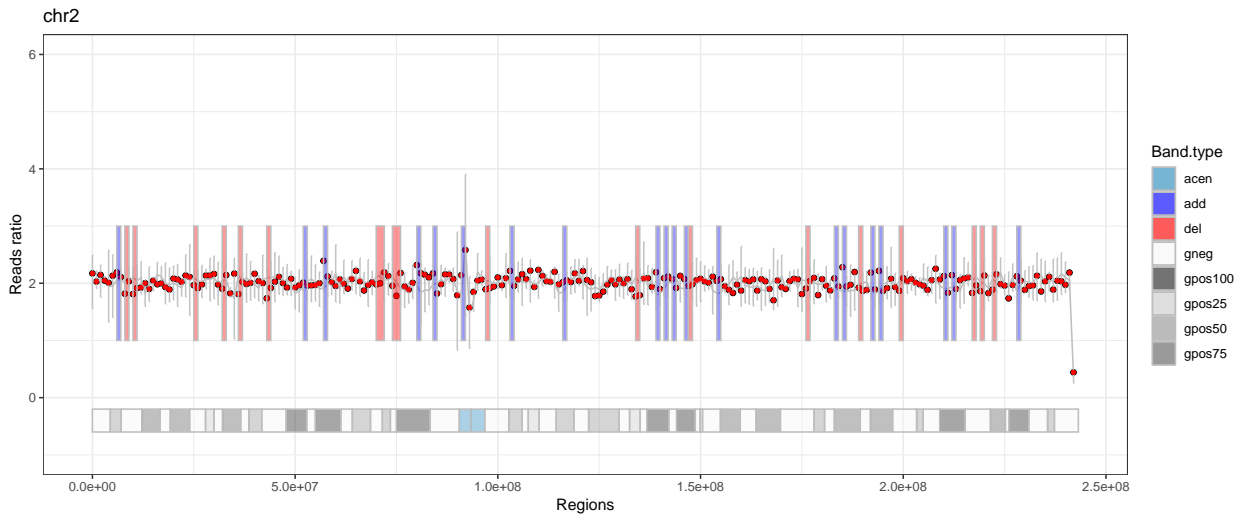
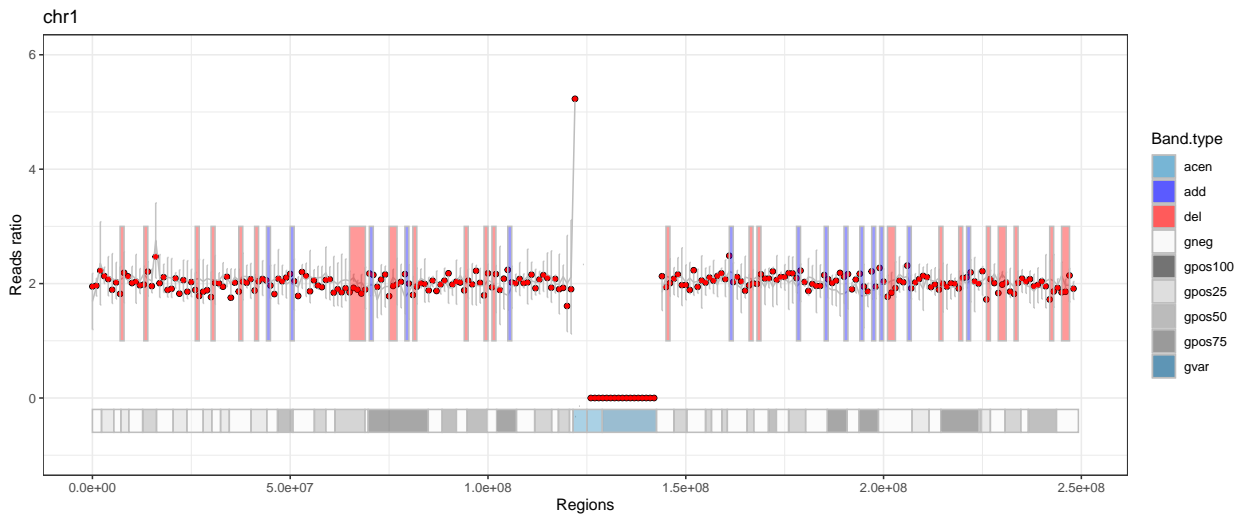
Bailey et al. (2018): Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell 173 (2), 371-385.e18. DOI: 10.1016/j.cell.2018.02.060.;

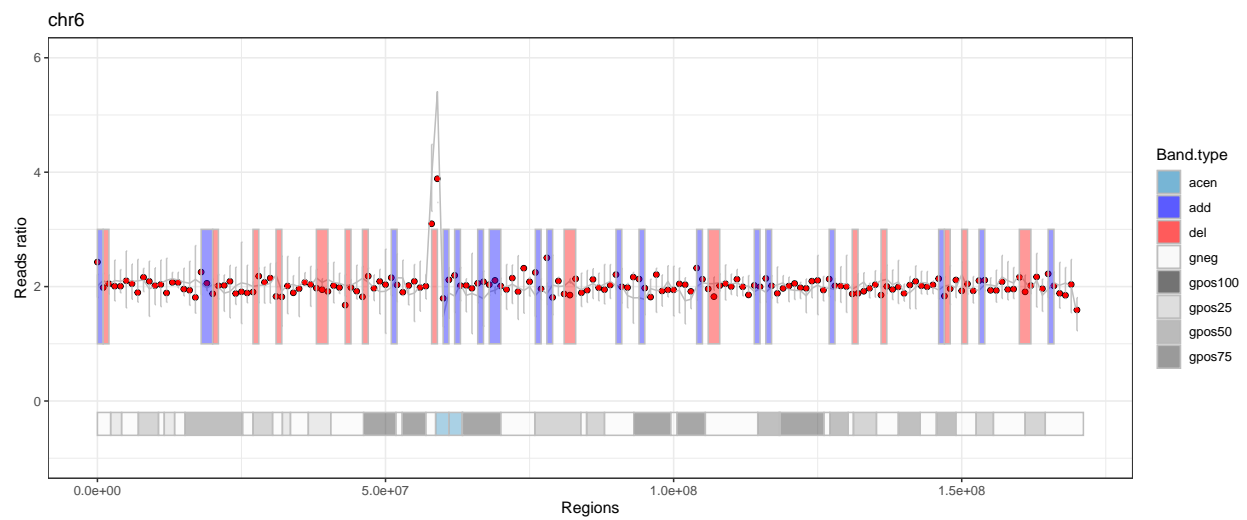
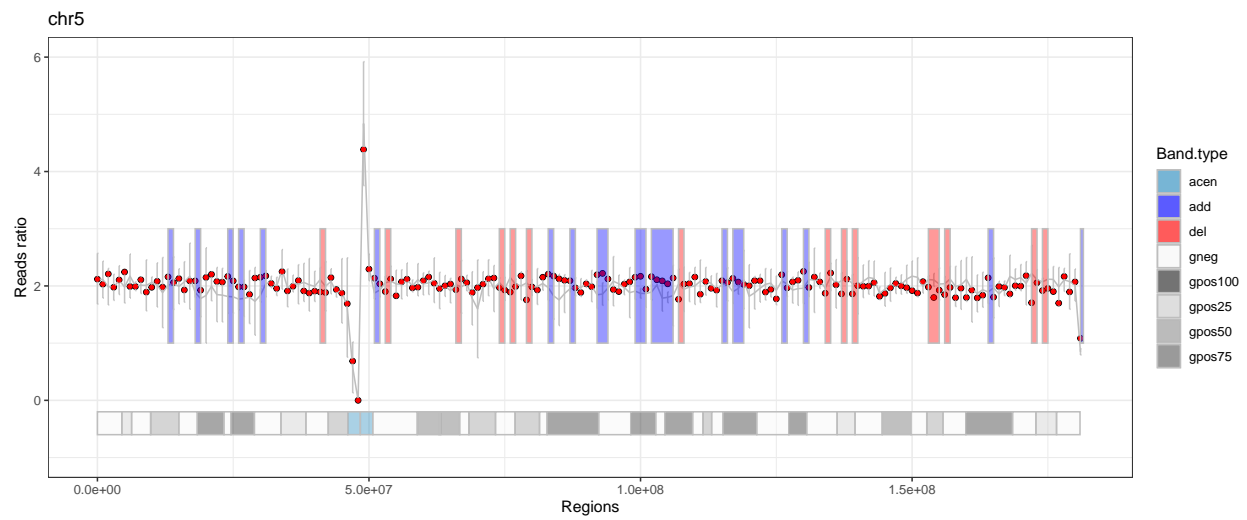
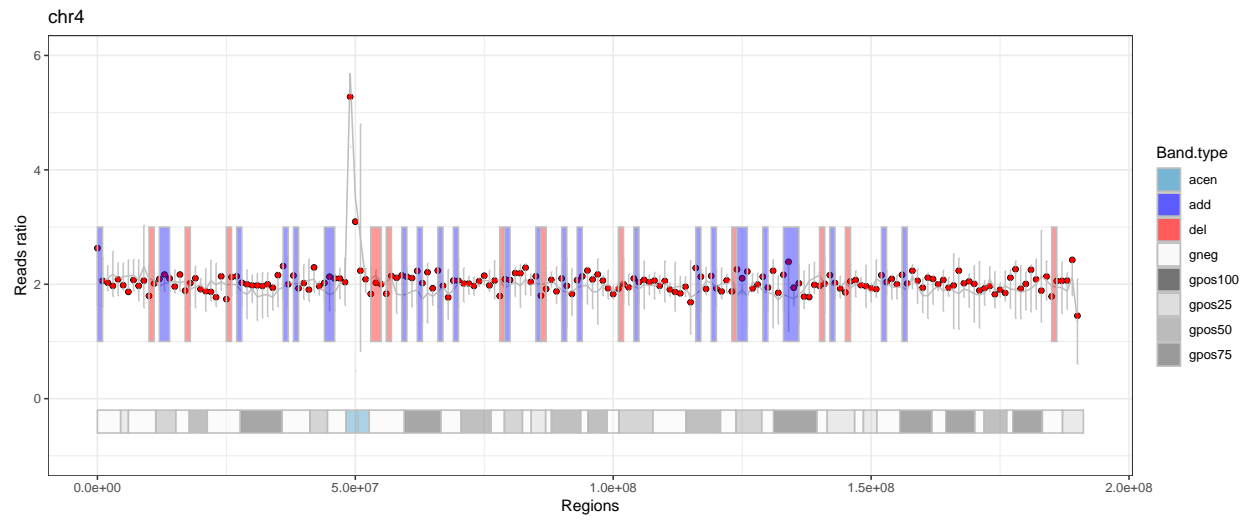
Papaemmanuil et al. (2016): Genomic Classification and Prognosis in Acute Myeloid Leukemia. The New England journal of medicine 374 (23), S. 2209–2221. DOI:10.1056/NEJMoa1516192.

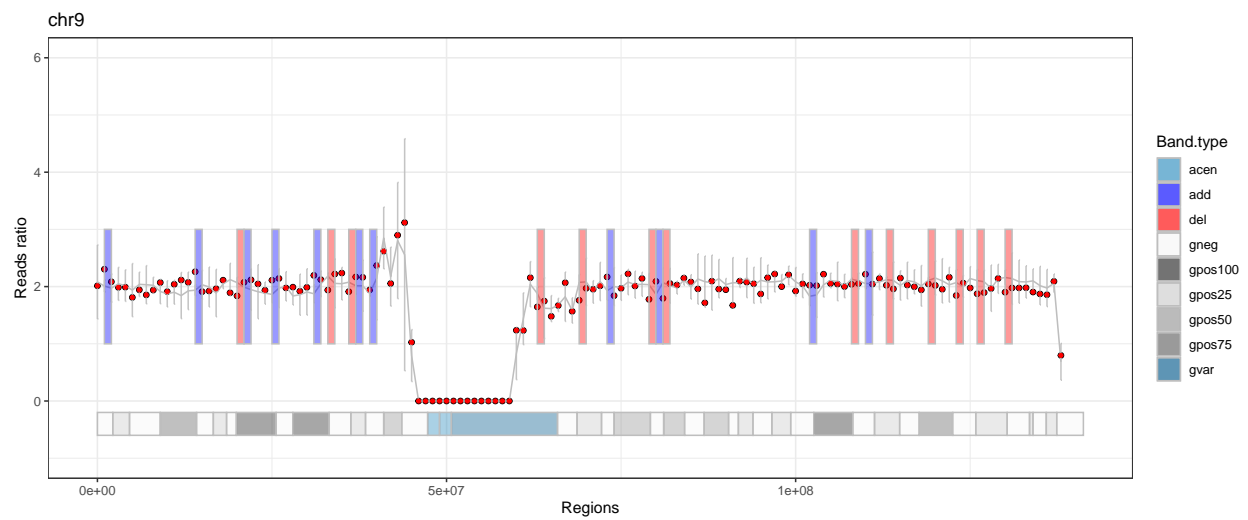
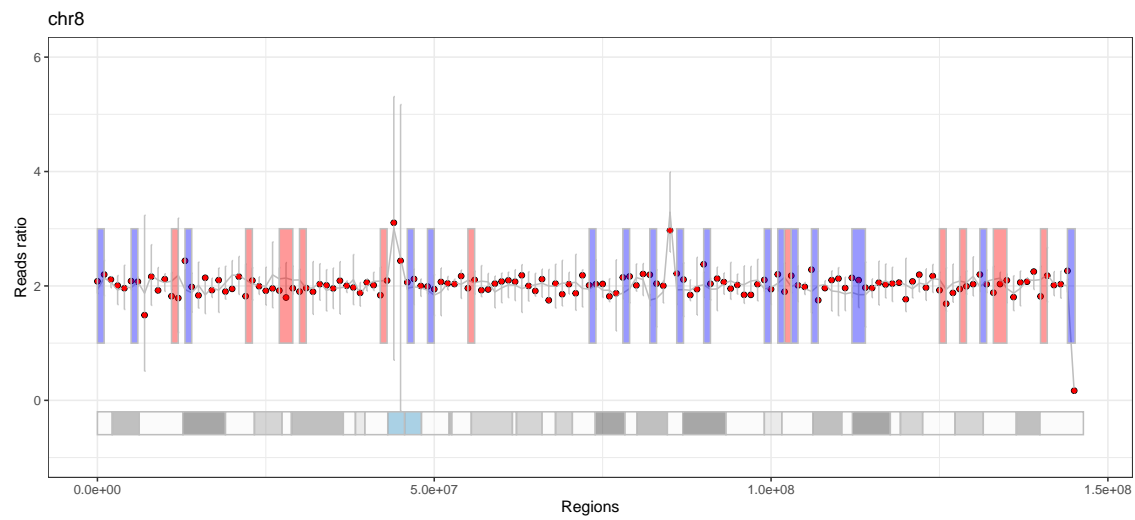
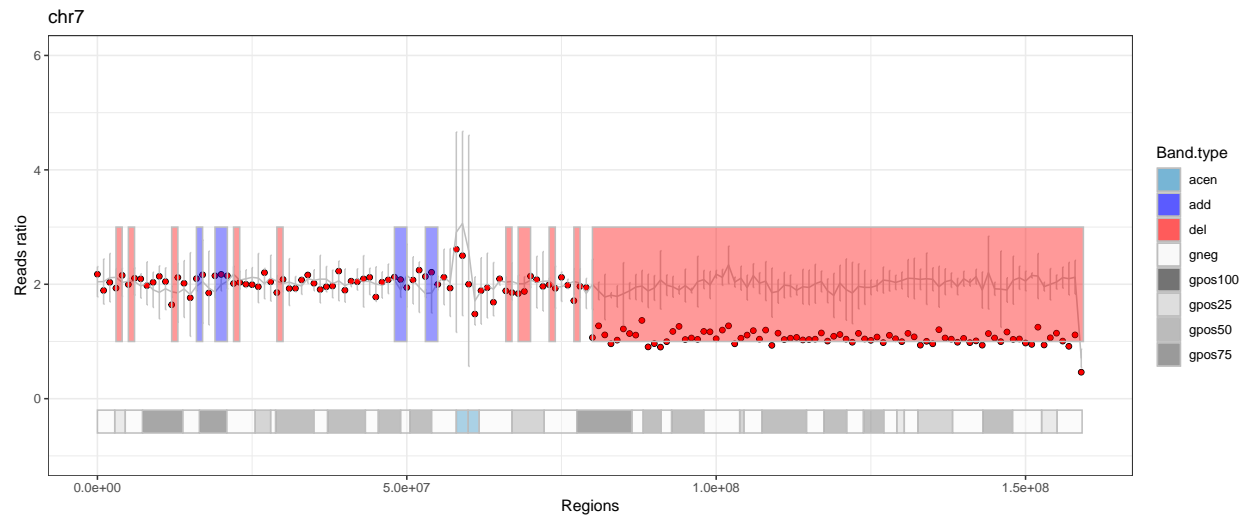
Karyotype overview:

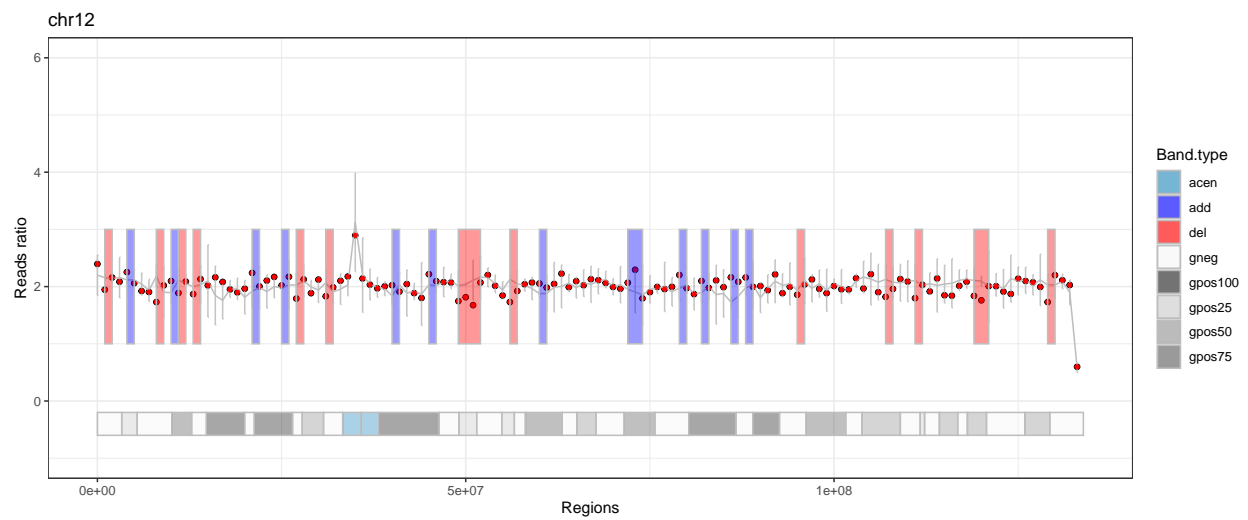
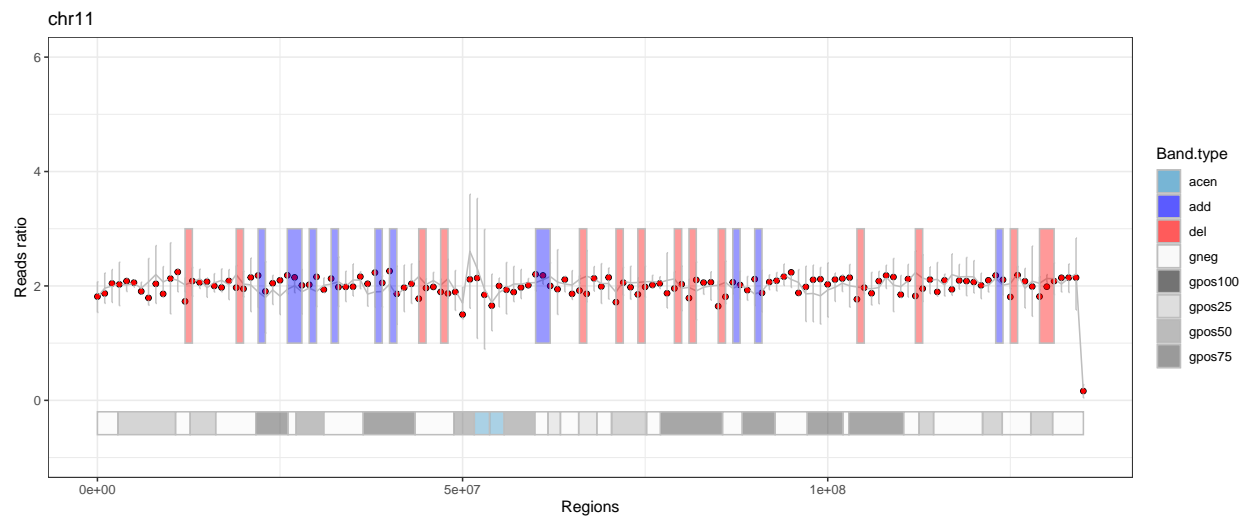
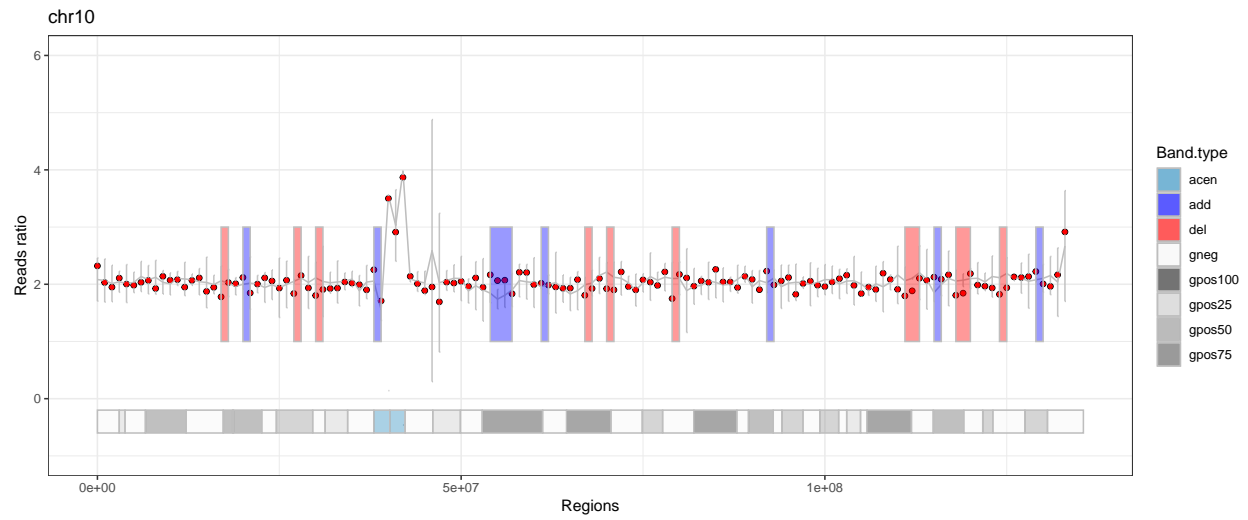


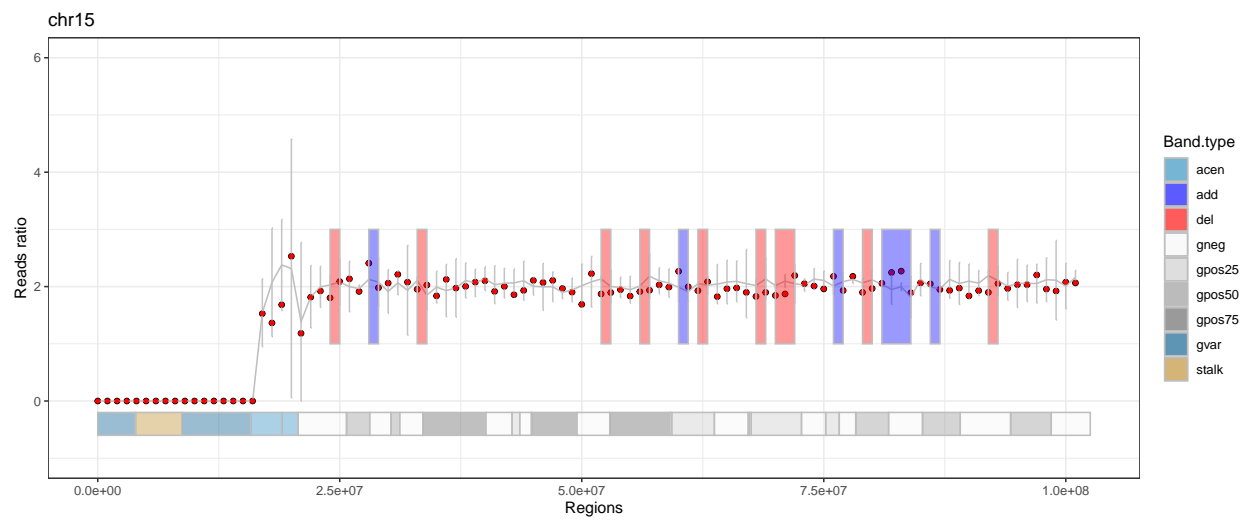
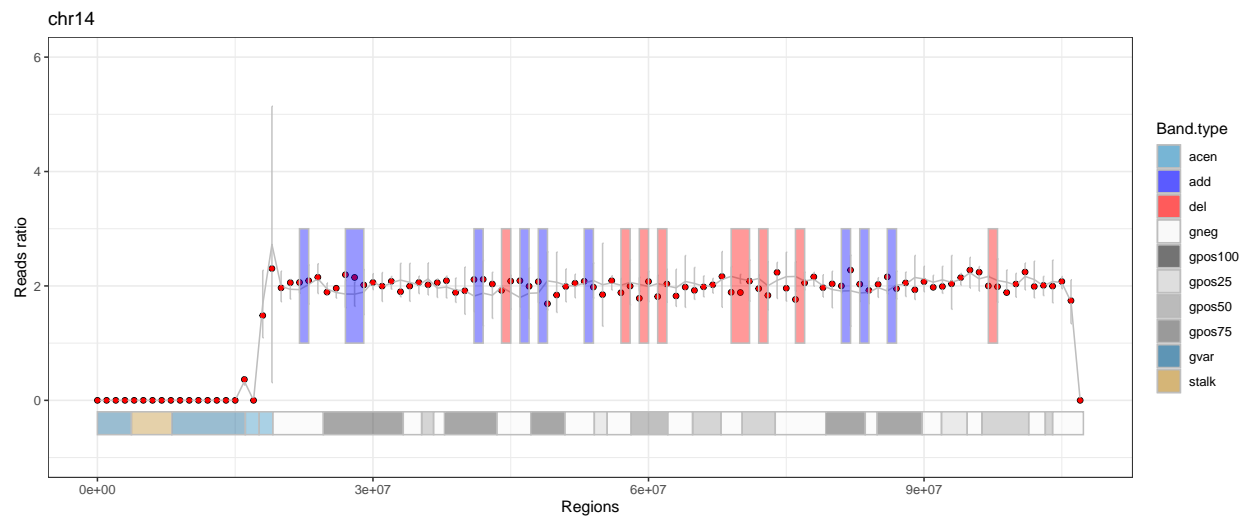
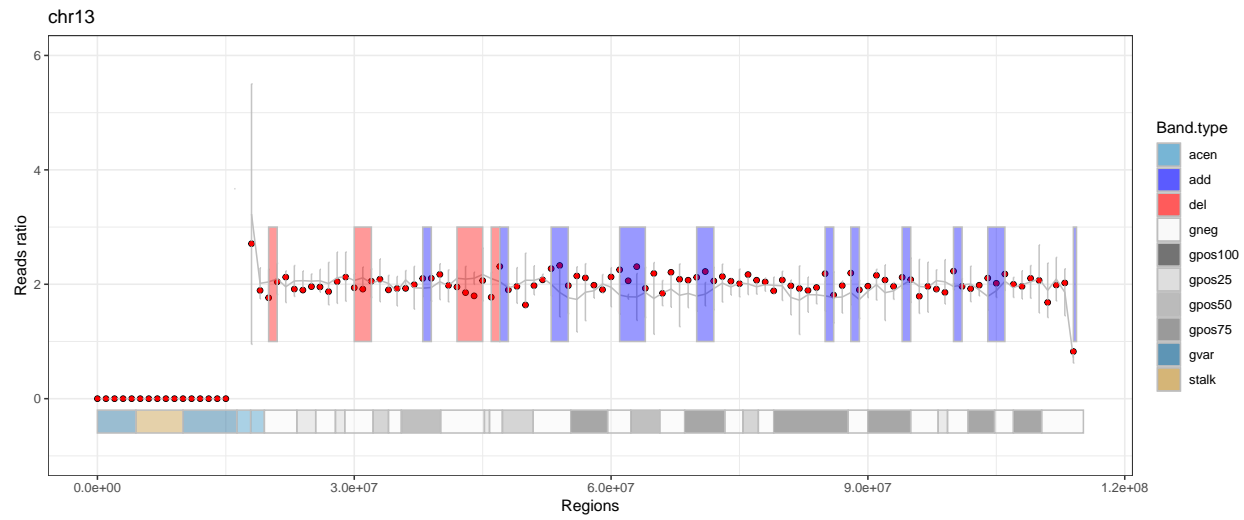
Chromosome Plots:

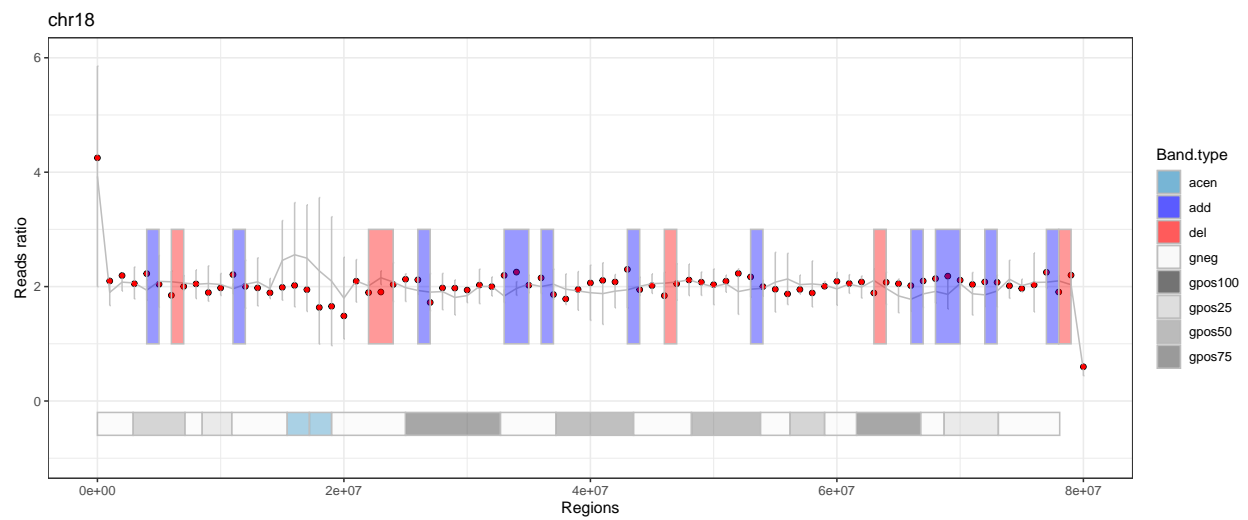
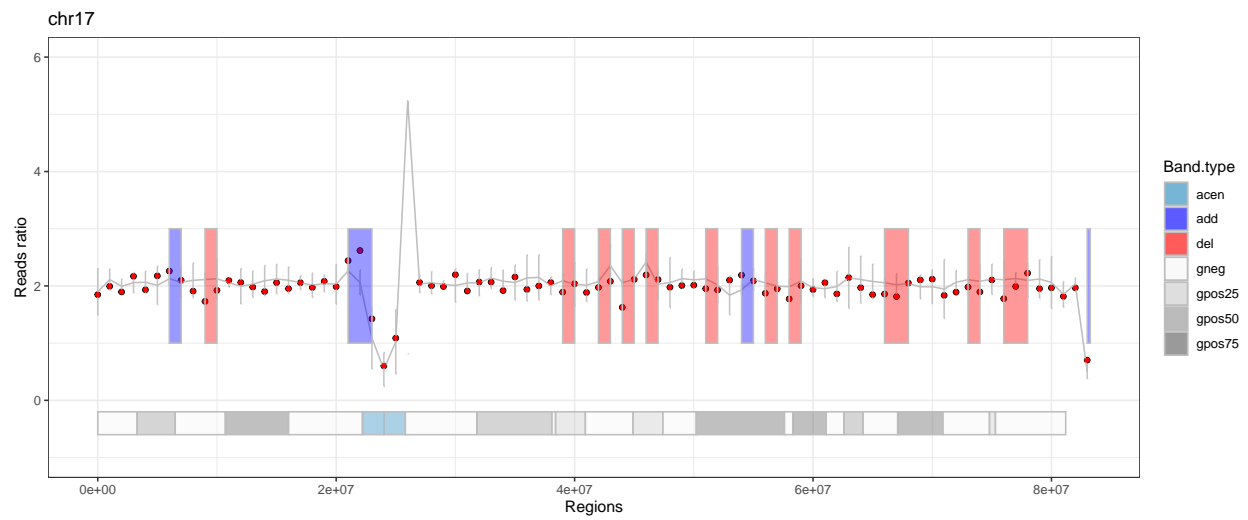
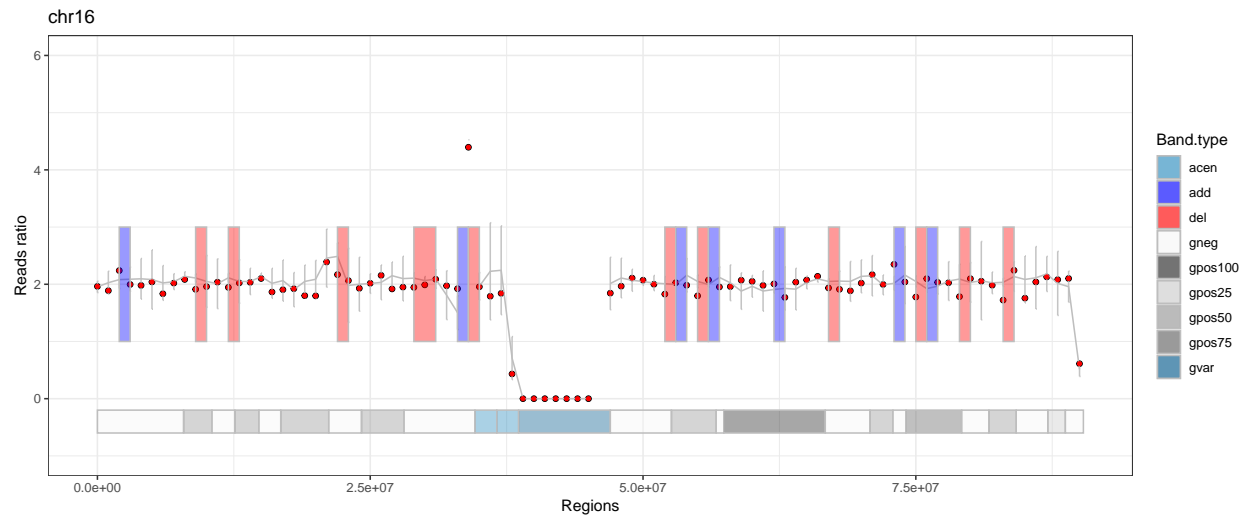


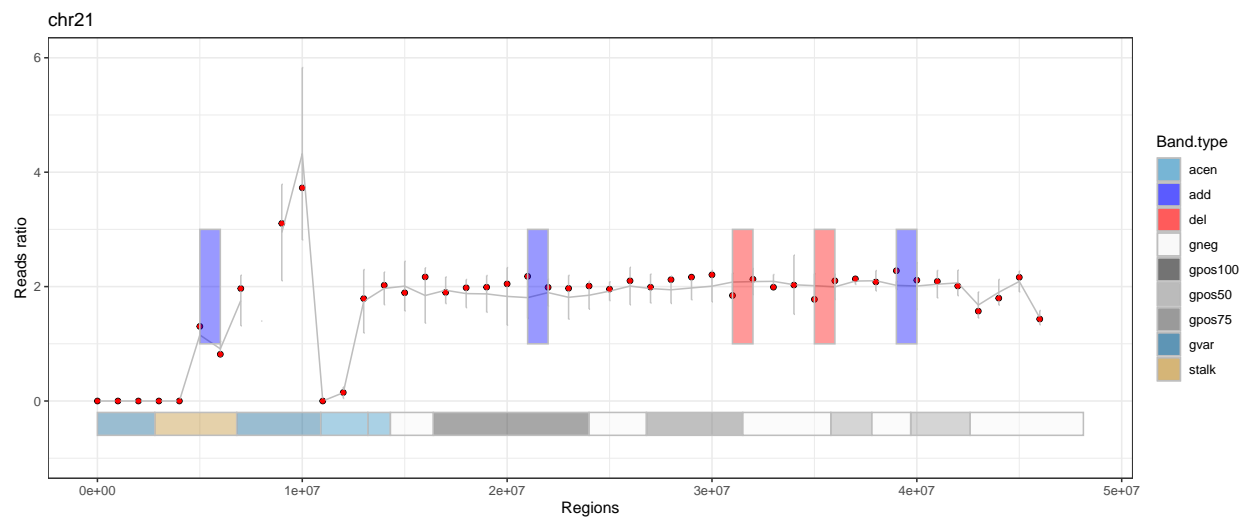
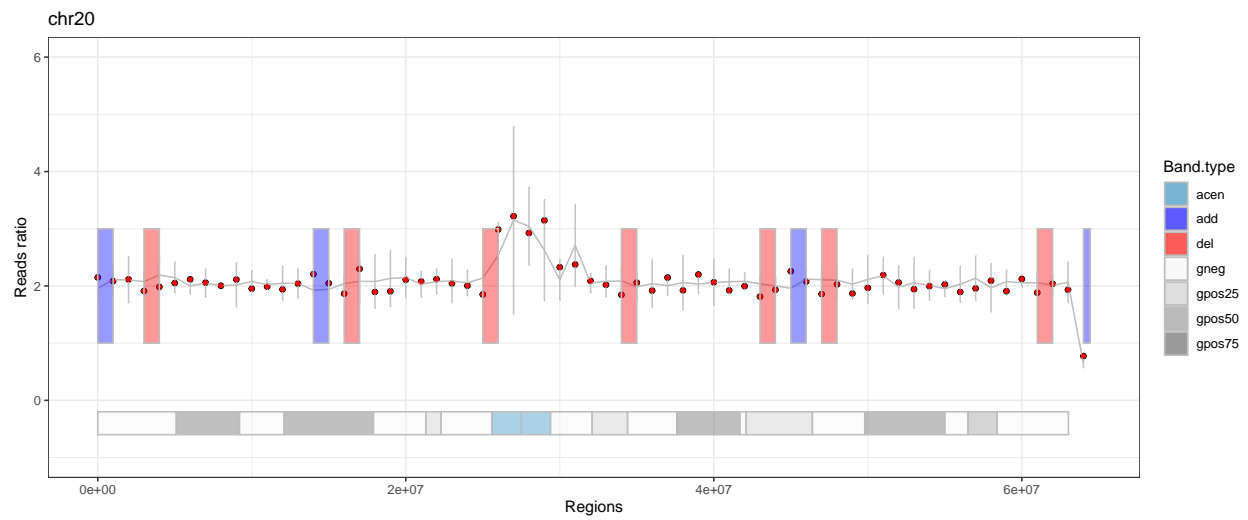
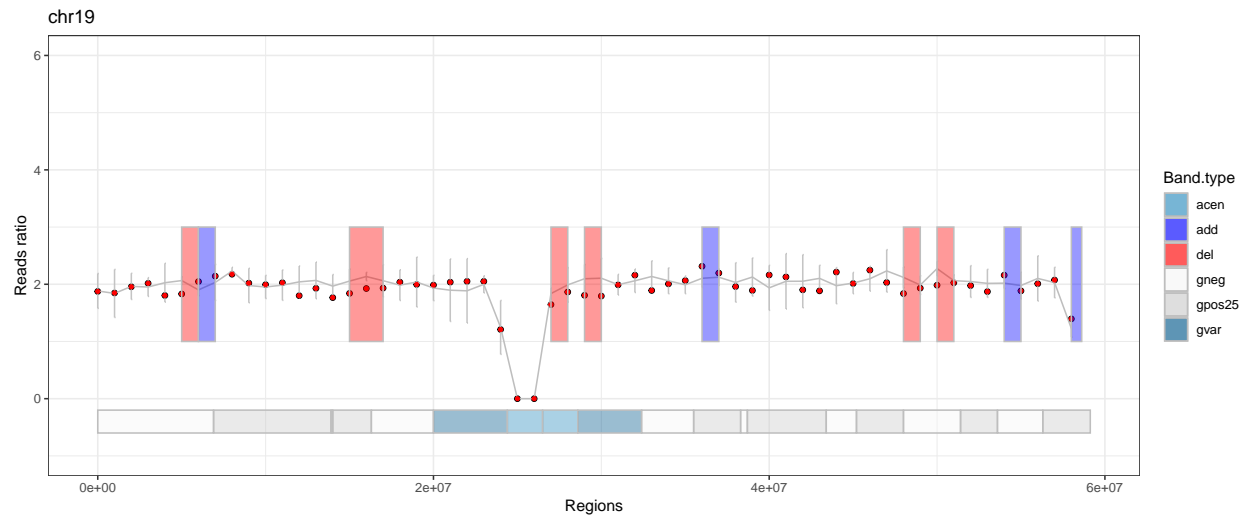


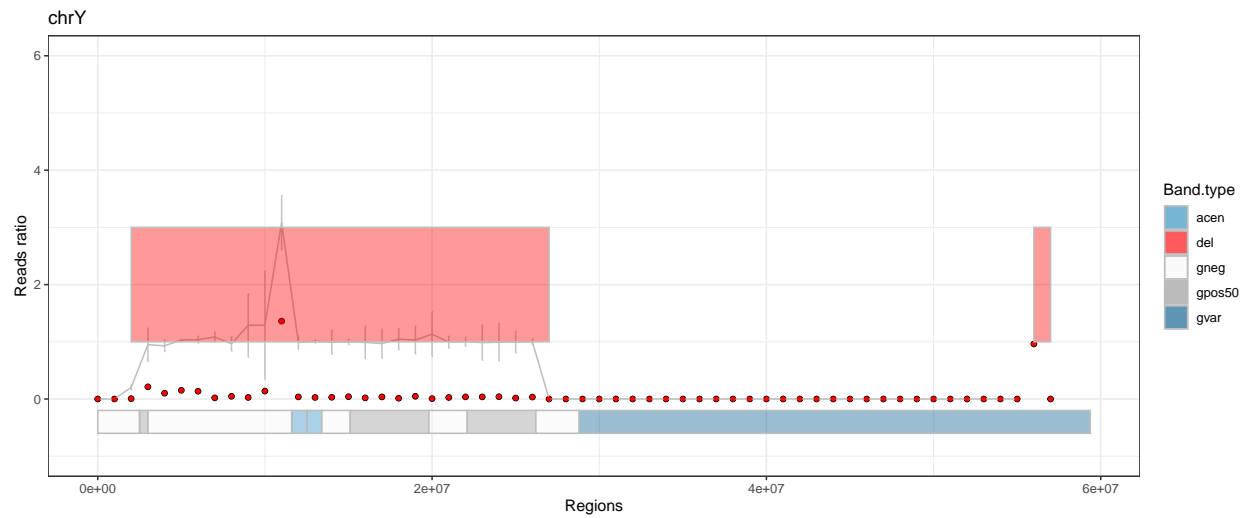
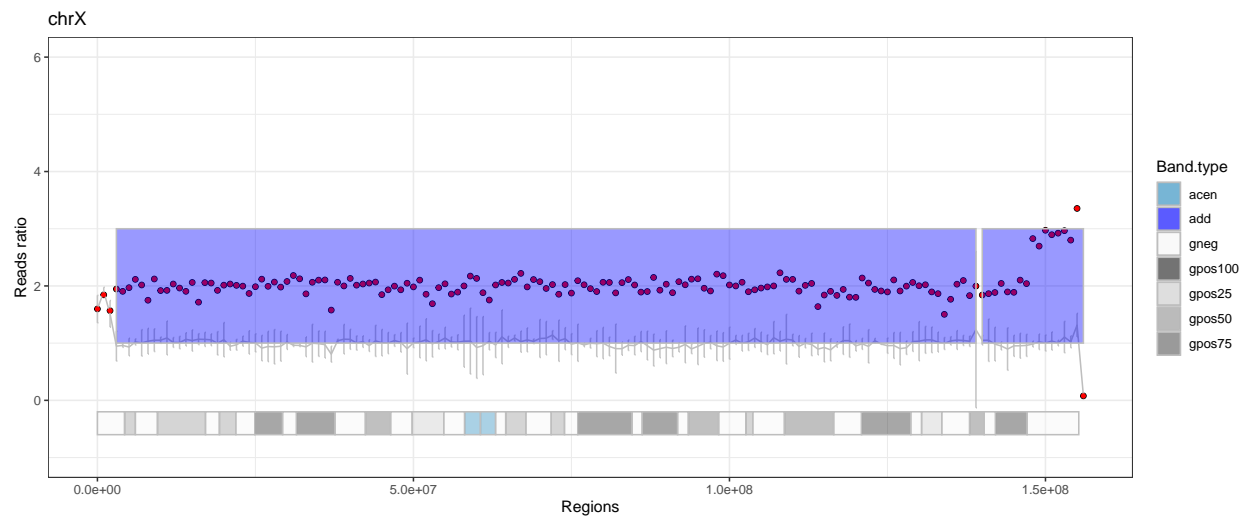
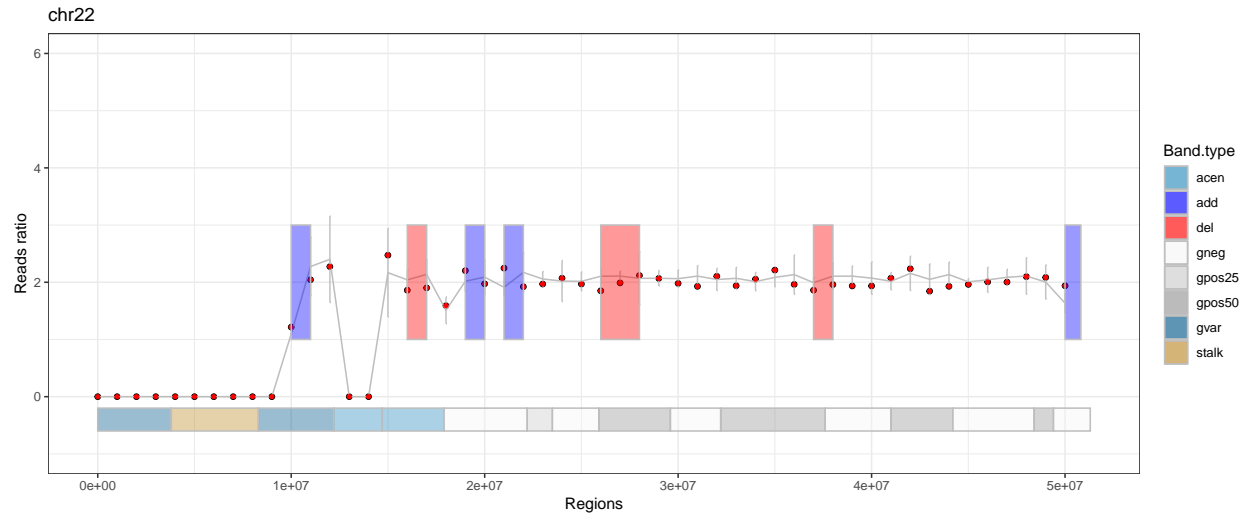












The legend contains two characteristics:

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

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