# Report CORIANDR: ChrOmosomal abeRration Identifier AND Reporter in R

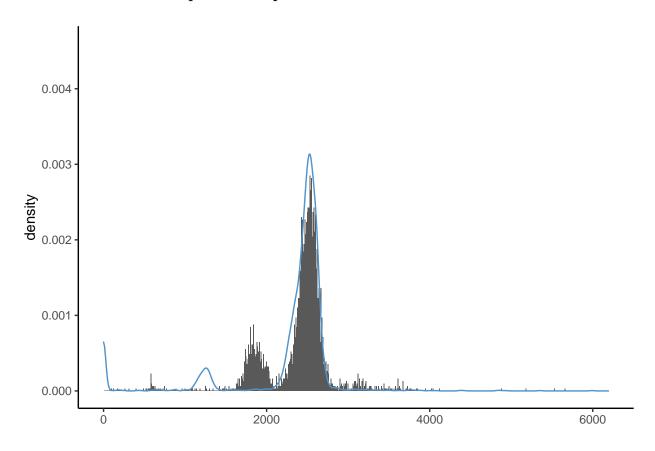
18 März, 2021

# Sample ID

## 109516.new.report

row read pairs average read length	3767290 $145.304$
unique mapped pairs	3617856
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)(q42.13q42.13)
- add(1)(q42.2q42.3)
- add(2)(p16.3p16.3)
- add(2)(q24.1q24.1)
- add(3)(p26.3p26.3)
- del(3)(p26.2p26.2)
- add(3)(q22.3q22.3)
- add(3)(q24q24)
- del(4)(q21.23q21.23)
- add(4)(q23q23)
- del(5)(q14.3q33.2)
- del(6)(p21.1p11.1)
- del(6)(q12q16.3)
- add(6)(q25.2q25.2)
- del(6)(q26q27)
- del(7)(p14.1p11.2)
- del(7)(q11.21q36.3)
- del(8)(p23.3p23.3)
- add(8)(q21.13q21.13)
- add(9)(p21.2p21.2)
- add(10)(q23.33q23.33)
- add(11)(p15.3p11.12)
- add(11)(q12.1q25)
- del(12)(p13.2p11.23)
- add(12)(p11.21p11.21)
- del(12)(p11.1p11.1)
- del(12)(q13.11q13.11)
- del(12)(q21.2q21.31)
- del(12)(q21.33q23.2)
- add(13)(q22.3q22.3)
- add(14)(q12q12)
- add(14)(q31.2q31.2)
- del(14)(q32.33q32.33)
- add(15)(q21.1q21.1)
- del(15)(q21.3q21.3)

add(15)(q22.2q22.2)

add(15)(q25.2q25.2)

add(16)(q12.2q13)

del(16)(q24.2q24.2)

del(17)(q21.32q21.32)

add(17)(q22q23.1)

del(18)(p11.31q23)

del(19)(p13.3p13.3)

del(19)(q13.33q13.33)

del(20)(p12.3p12.3)

add(20)(q13.12q13.12)

add(21)(q22.12q22.12)

del(21)(q22.3q22.3)

del(22)(q11.21q11.21)

del(22)(q12.2q12.2)

add(X)(p22.33p11.21)

add(X)(q11.2q26.3)

add(X)(q27.2q28)

del(Y)(p11.31q11.23)

# Genes affected by CNVs\*:

id	sym	bol chr	sta	rt end	abe	rration
1604	ENSG00000123080	CDKN2C	chr1	50960745	50974634	del
8082	ENSG00000121989	ACVR2A	chr2	147844517	147930826	amp
8662	ENSG00000064933	PMS1	chr2	189784085	189877629	$\operatorname{del}$
11346	ENSG00000181722	ZBTB20	chr3	114314501	115147271	$\operatorname{del}$
13641	ENSG00000134853	PDGFRA	chr4	54229280	54298245	del
13646	ENSG00000157404	KIT	chr4	54657918	54740715	del
14974	ENSG00000109670	FBXW7	chr4	152320544	152536092	del
16825	ENSG00000145715	RASA1	chr5	87267888	87391931	del
17123	ENSG00000134982	APC	chr5	112707498	112846239	del
17886	ENSG00000182578	CSF1R	chr5	150053291	150113372	del
17889	ENSG00000113721	PDGFRB	chr5	150113839	150155872	del
19585	ENSG00000124762	CDKN1A	chr6	36676460	36687339	del
21384	ENSG00000085511	MAP3K4	chr6	160991727	161117385	amp
21681	ENSG00000122512	PMS2	chr7	5970925	6009106	del
22481	ENSG00000146648	EGFR	chr7	55019017	55211628	del
22928	ENSG00000263001	GTF2I	chr7	74650231	74760692	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

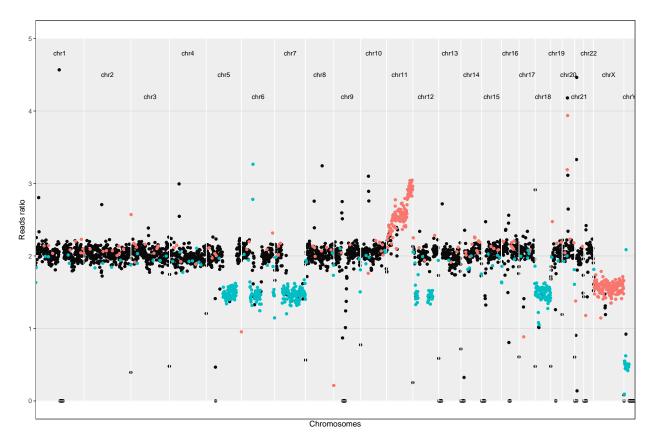
id	sym	bol chr	sta	rt end	abe	rration
24082	ENSG00000157764	BRAF	chr7	140719327	140924928	del
24217	ENSG00000197993	KEL	chr7	142941114	142962363	$\operatorname{del}$
24328	ENSG00000055130	CUL1	chr7	148697914	148801110	del
24329	ENSG00000106462	EZH2	chr7	148807383	148884321	del
24436	ENSG00000106615	RHEB	chr7	151466012	151520120	del
24449	ENSG00000055609	KMT2C	chr7	152134922	152436005	del
31516	ENSG00000097007	ABL1	chr9	130713946	130887675	del
31674	ENSG00000148400	NOTCH1	chr9	136494433	136546048	del
32366	ENSG00000133818	RRAS2	chr11	14277922	14364506	amp
32621	ENSG00000184937	WT1	chr11	32387775	32435885	amp
33142	ENSG00000198561	CTNND1	chr11	57753243	57819546	amp
33487	ENSG00000168066	SF1	chr11	64764606	64778786	amp
33490	ENSG00000133895	MEN1	chr11	64803510	64811294	amp
33764	ENSG00000110092	CCND1	chr11	69641087	69654474	amp
33855	ENSG00000165458	INPPL1	chr11	72223701	72239147	amp
34405	ENSG00000082175	PGR	chr11	101029624	101130524	amp
34521	ENSG00000149311	ATM	chr11	108222484	108369102	amp
34754	ENSG00000118058	KMT2A	chr11	118436464	118526832	amp
35859	ENSG000001165731	RET	chr10	43077069	43130351	del
37122	ENSG00000148737	TCF7L2	chr10	112950247	113167678	amp
37950	ENSG00000111276	CDKN1B	chr12	12715058	12722369	del
37989	ENSG00000171270	ATF7IP	chr12	14365632	14502935	del
38141	ENSG00000171001	KRAS	chr12	25204789	25250936	del
39959	ENSG00000179295	PTPN11	chr12	112418351	112509913	amp
45273	ENSG00000119250	TCF12	chr15	56918623	57299281	del
46467	ENSG00000110202 ENSG00000103126	AXIN1	chr16	287440	352723	del
46622	ENSG00000103197	TSC2	chr16	2047967	2089491	del
47953	ENSG00000166164	BRD7	chr16	50313487	50368934	amp
47966	ENSG00000180101	CYLD	chr16	50742050	50801935	amp
48110	ENSG000000009199	NUP93	chr16	56730118	56850286	amp
48288	ENSG00000162966	CBFB	chr16	67028984	67101058	del
48327	ENSG000000102974	CTCF	chr16	67562467	67639177	del
50883	ENSG00000102971 ENSG00000120071	KANSL1	chr17	46029916	46225389	del
52042	ENSG00000120071	ZNF750	chr17	82829434	82840022	del
52137	ENSG00000177426	TGIF1	chr18	3411608	3459978	del
52741	ENSG00000177120	SETBP1	chr18	44680173	45068510	del
52799	ENSG00000175387	SMAD2	chr18	47808957	47931146	del
52876	ENSG00000110501	SMAD4	chr18	51028394	51085045	del
53051	ENSG00000171791	BCL2	chr18	63123346	63320128	del
247031	ENSG00000171731	TMSB4X	chrX	12975110	12977227	amp
248091	ENSG00000173674	EIF1AX	chrX	20124525	20141838	amp
248131	ENSG00000177189	RPS6KA3	chrX	20149911	20267519	amp
249381	ENSG00000177163 ENSG00000198947	DMD	chrX	31097677	33339441	amp
250291	ENSG00000136317 ENSG00000183337	BCOR	chrX	40049815	40177329	amp
250501	ENSG00000124486	USP9X	chrX	41085445	41236579	amp
250551	ENSG00000124400 ENSG00000215301	DDX3X	chrX	41333284	41364472	amp
250001 $251001$	ENSG00000213301 ENSG00000147050	KDM6A	chrX	44873177	45112602	amp
251001 $251431$	ENSG00000147030 ENSG00000182872	RBM10	chrX	47145221	47186813	amp
251431 $251601$	ENSG00000132872 ENSG00000078061	ARAF	chrX	47561100	47571920	amp
251001 $253891$	ENSG00000126012	KDM5C	chrX	53191321	53225422	
253981 $253981$	ENSG00000120012 ENSG00000072501	SMC1A	chrX	53374149	53223422 $53422728$	amp
253961 $254051$	ENSG00000072301 ENSG000000086758	HUWE1	chrX	53532096	53686728	amp
204001	TUDG00000000100	110 11 11	OHA	55552U3U	00000120	ашр

id	sym	bol chr	$\operatorname{sta}$	rt end	abe	rration
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

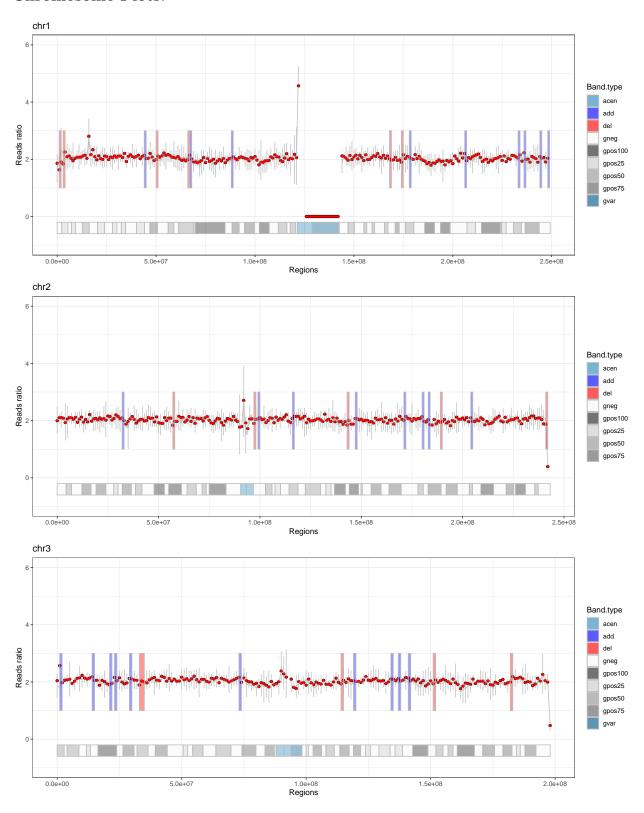
Bailey et al. (2018): Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell 173 (2), 371-385.e18. DOI: 10.1016/j.cell.2018.02.060.;

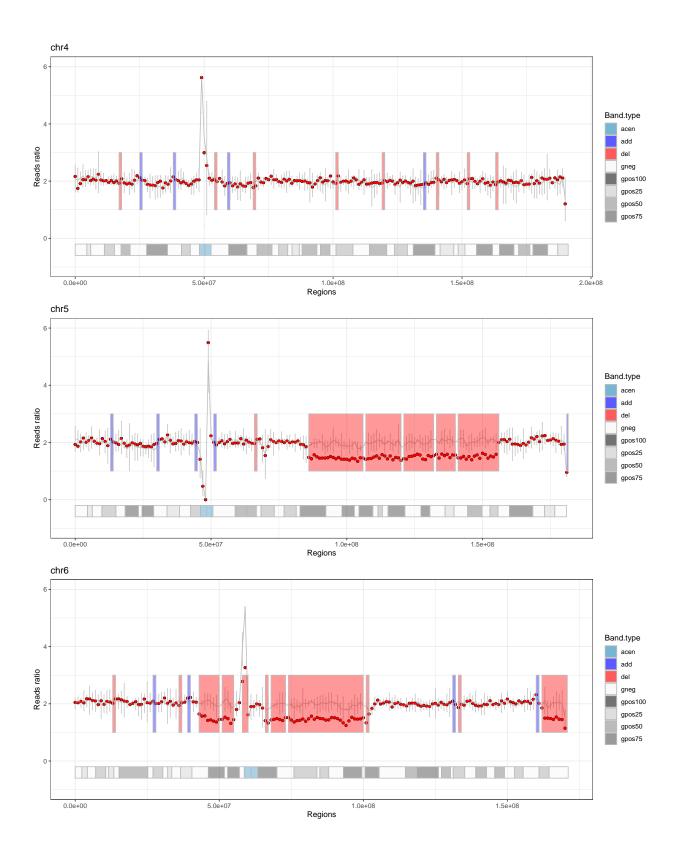
Papaemmanuil et al. (2016): Genomic Classification and Prognosis in Acute Myeloid Leukemia. The New England journal of medicine 374 (23), S. 2209–2221. DOI:10.1056/NEJMoa1516192.

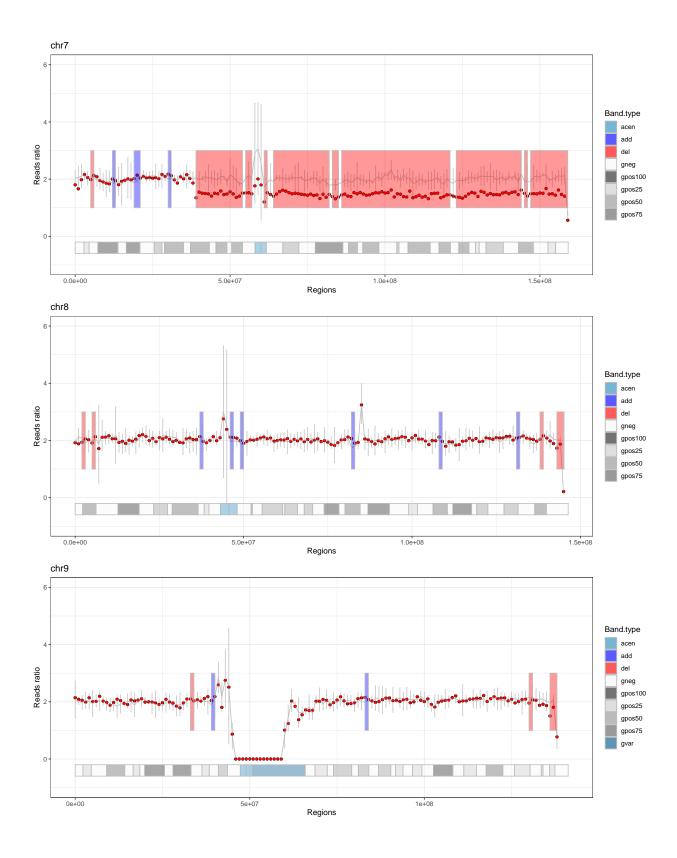
# Karyotype overview:

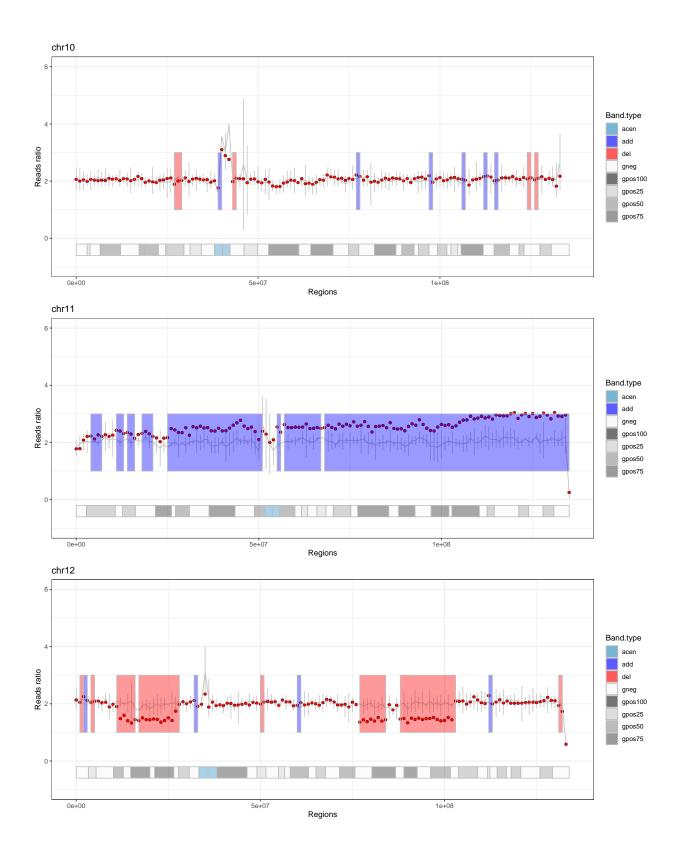


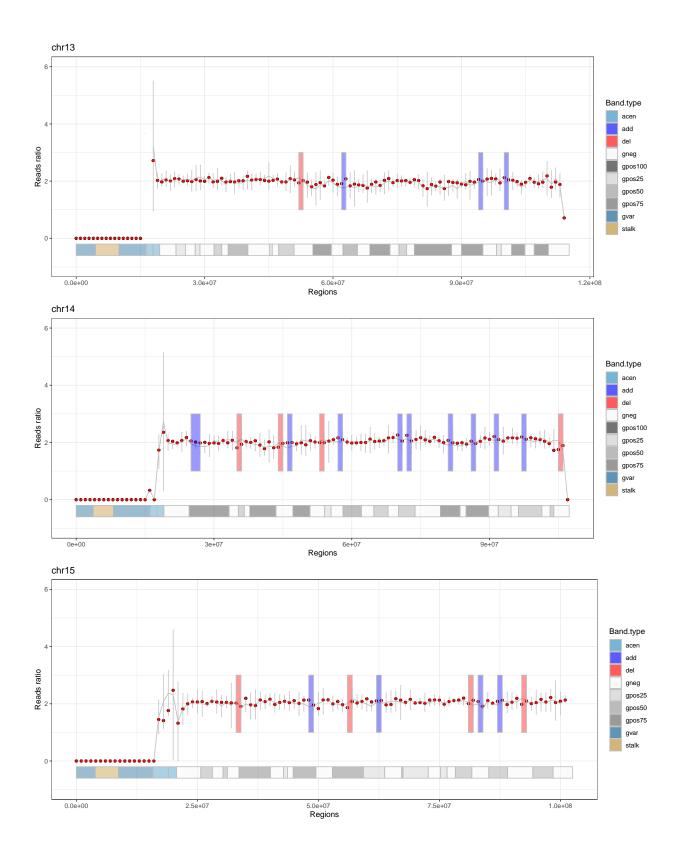
## **Chromosome Plots:**

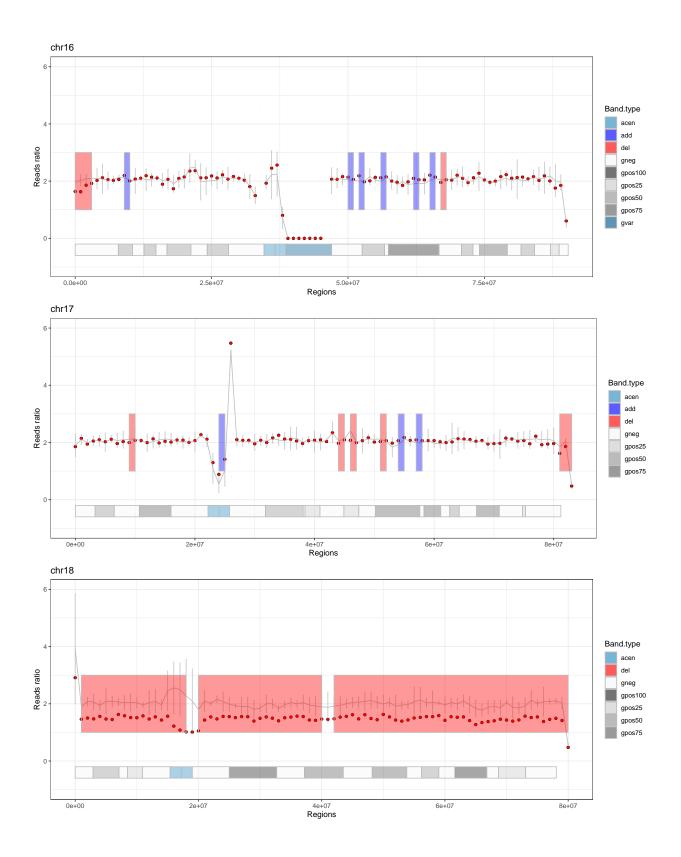


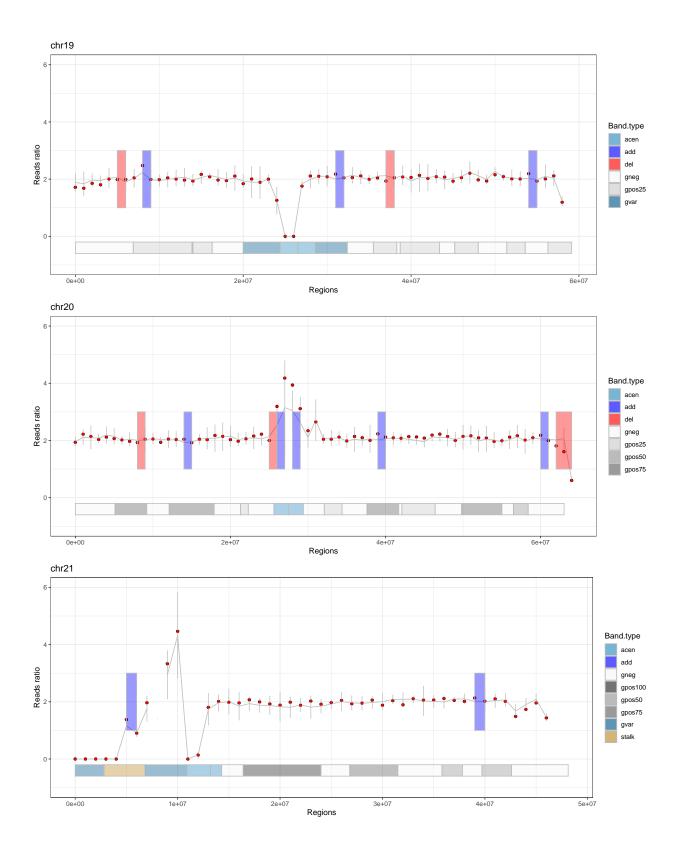


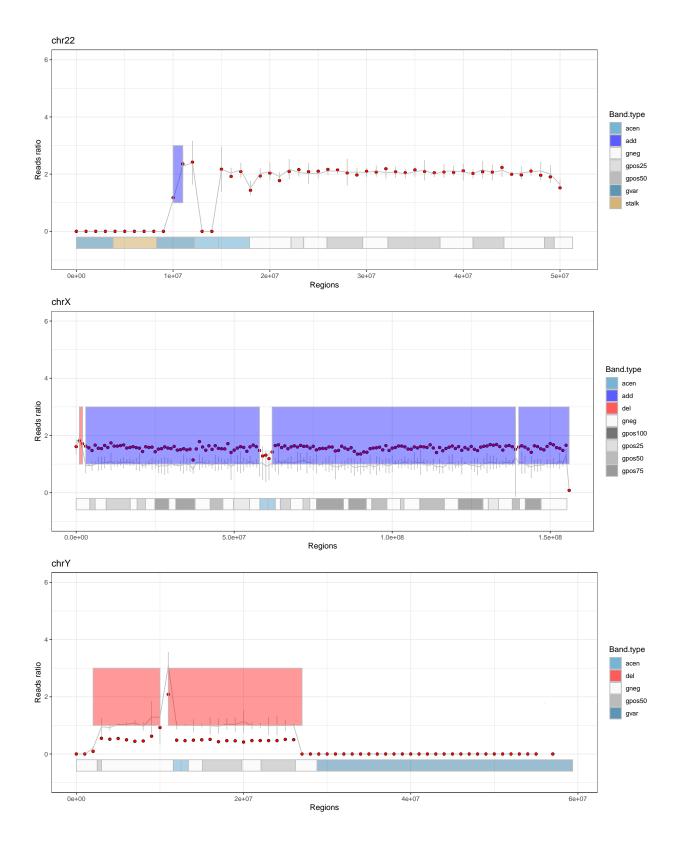












#### The legend contains two characteristics:

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