Report CORIANDR: ChrOmosomal abeRration Identifier AND Reporter in R

09 Dezember, 2020

| unique_reads | 1922410 |
|-------------------------|-----------------------|
| $average_length$ | 149.5 |
| $unique_mapped_reads$ | 3693021 |
| SAMPLE_ID | $120517.\mathrm{new}$ |
| gender | M |

Calculated numerical karyotype:

add(1)(q25.3q25.3)

add(1)(q43q43)

add(2)(p24.3p24.3)

add(2)(p24.1p24.1)

add(2)(p11.2p11.2)

del(2)(q11.1q37.3)

add(3)(p14.1p14.1)

del(3)(q22.1q22.3)

add(4)(p11p11)

add(4)(q35.2q35.2)

add(5)(p15.33p15.33)

add(5)(p15.31p15.2)

 $\mathrm{add}(5)(\mathrm{p}13.2\mathrm{p}13.2)$

add(5)(q12.1q12.1)

del(5)(q15q23.1)

del(5)(q23.3q31.1)

add(5)(q31.3q32)

del(5)(q33.1q33.3)

add(5)(q34q35.1)

del(6)(p22.3p22.3)

del(6)(q13q27)

add(7)(p12.1p12.1)

add(7)(q31.32q31.32)

del(8)(p23.3p11.22)

add(8)(p11.21q24.3)

- add(9)(p24.3p24.3)
- add(9)(p21.3p21.3)
- add(9)(q31.1q31.2)
- del(9)(q33.3q34.3)
- add(10)(p11.21p11.21)
- add(10)(q11.1q11.1)
- ${\rm add}(10)(q11.23q11.23)$
- add(10)(q24.33q25.1)
- add(10)(q26.3q26.3)
- del(11)(p15.5p13)
- add(11)(p11.12q12.1)
- add(11)(q14.1q14.1)
- add(11)(q21q21)
- add(11)(q24.2q24.2)
- del(12)(p13.2p11.21)
- add(13)(p11.1q12.3)
- del(13)(q13.1q34)
- add(14)(p11.1q11.1)
- add(14)(q31.3q31.3)
- add(15)(q12q13.1)
- add(15)(q14q14)
- del(16)(p11.2p11.1)
- del(16)(q11.2q24.3)
- del(17)(p13.3p12)
- del(18)(p11.31p11.1)
- del(18)(q11.2q23)
- del(20)(p13p11.21)
- add(20)(q11.21q11.22)
- del(20)(q11.23q13.33)
- add(21)(p12p11.1)
- add(21)(q21.3q21.3)
- ${\rm add}(22)({\rm p}11.2{\rm q}12.2)$
- del(22)(q12.3q12.3)

Genes affected by CNVs*:

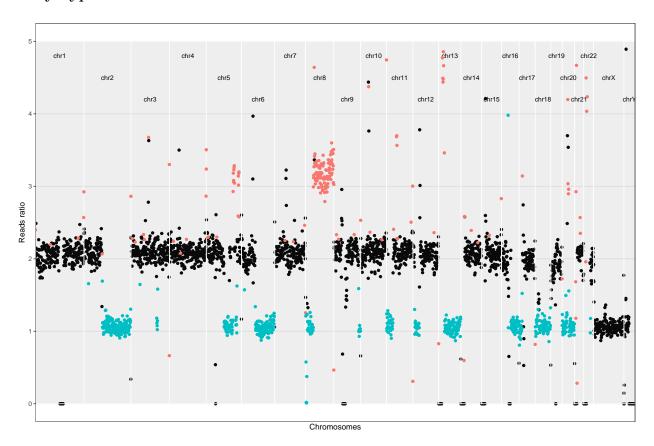
| id | sym | bol chr | sta | rt end | abe | rration |
|----------------|------------------------------------|----------------|---|-----------|-----------|----------------------|
| 7505 | ENSG00000153094 | BCL2L11 | chr2 | 111119378 | 111168445 | del |
| 8082 | ENSG00000121989 | ACVR2A | chr2 | 147844517 | 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 |
| 11808 | ENSG00000161382 | PIK3CB | chr3 | 138652699 | 138834938 | del |
| 11814 | ENSG00000031362 ENSG00000183770 | FOXL2 | chr3 | 138944224 | 138947137 | del |
| 17123 | ENSG00000134982 | APC | $\frac{\mathrm{chr}5}{\mathrm{chr}5}$ | 112707498 | 112846239 | del |
| 20143 | ENSG00000154502 ENSG00000156508 | EEF1A1 | $\frac{\mathrm{chr}_{6}}{\mathrm{chr}_{6}}$ | 73515750 | 73523797 | del |
| 20743 | ENSG00000130308 ENSG00000047936 | ROS1 | $\frac{\mathrm{chr}_{6}}{\mathrm{chr}_{6}}$ | 117288300 | 117425855 | del |
| 20713 21001 | ENSG00000047930 ENSG00000118503 | TNFAIP3 | $\frac{\mathrm{cm}\sigma}{\mathrm{chr}6}$ | 137867214 | 137883312 | del |
| 21164 | ENSG00000118303 ENSG00000131023 | LATS1 | $\frac{\mathrm{cm}\sigma}{\mathrm{chr}6}$ | 149658153 | 149718105 | del |
| 21104 21223 | ENSG00000131023 ENSG00000091831 | ESR1 | $\frac{\mathrm{cm}\sigma}{\mathrm{chr}6}$ | 151656691 | 152129619 | del |
| 21223 | ENSG00000091831 ENSG00000085511 | MAP3K4 | chr6 | 160991727 | 161117385 | del |
| 27499 | ENSG00000083311 ENSG00000179388 | EGR3 | chr8 | 22687659 | 22693480 | del |
| 27499 27807 | ENSG00000179388 ENSG00000077782 | FGFR1 | chr8 | 38400215 | 38468834 | del |
| 21001 | ENSG00000017782 ENSG00000164736 | SOX17 | chr8 | 54457935 | 54460892 | |
| 28554 | | CNBD1 | chr8 | 86866442 | 87615219 | amp |
| 28965 | ENSG00000176571 | RAD21 | | 116845935 | | amp |
| | ENSG00000164754 | | chr8 | | 116874866 | amp |
| 29133 | ENSG00000136997 | MYC CDTA N1 | chr8 | 127735434 | 127742951 | amp |
| 31438 | ENSG00000197694 | SPTAN1 | chr9 | 128552558 | 128633662 | del |
| 31516 | ENSG00000097007 ENSG00000165699 | ABL1 TSC1 | chr9 chr9 | 130713946 | 130887675 | del |
| 31558 | | | | 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 |
| 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 |
| 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 |
| 47953 | ENSG00000166164 | BRD7 | chr16 | 50313487 | 50368934 | del |
| | | | | | | |

| id | sym | bol chr | sta | rt end | abe | rration |
|-------|-----------------|---------|----------------------|----------|----------|----------------------|
| 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 |
| 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 |
| 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 |

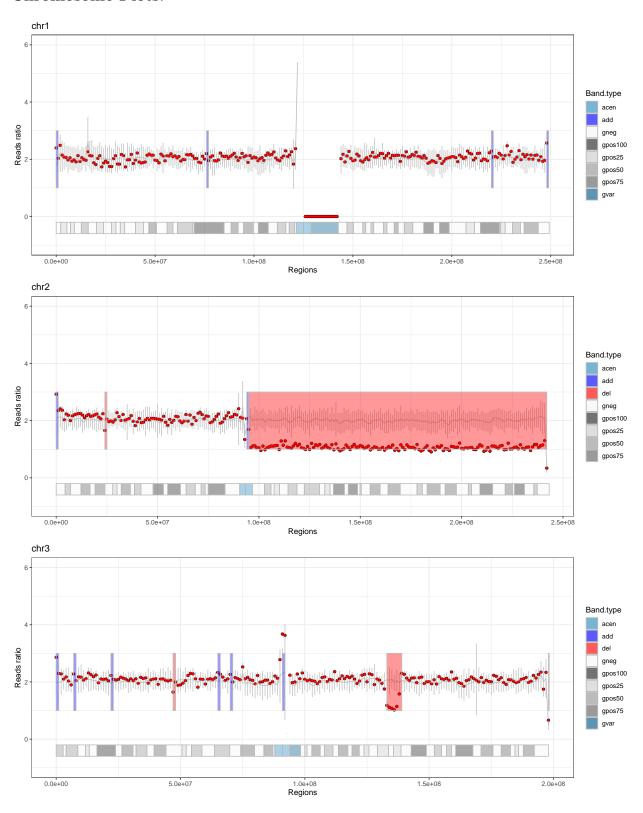
Bailey et al. (2018): Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell 173 (2), 371-385.e18. DOI: 10.1016/j.cell.2018.02.060.;

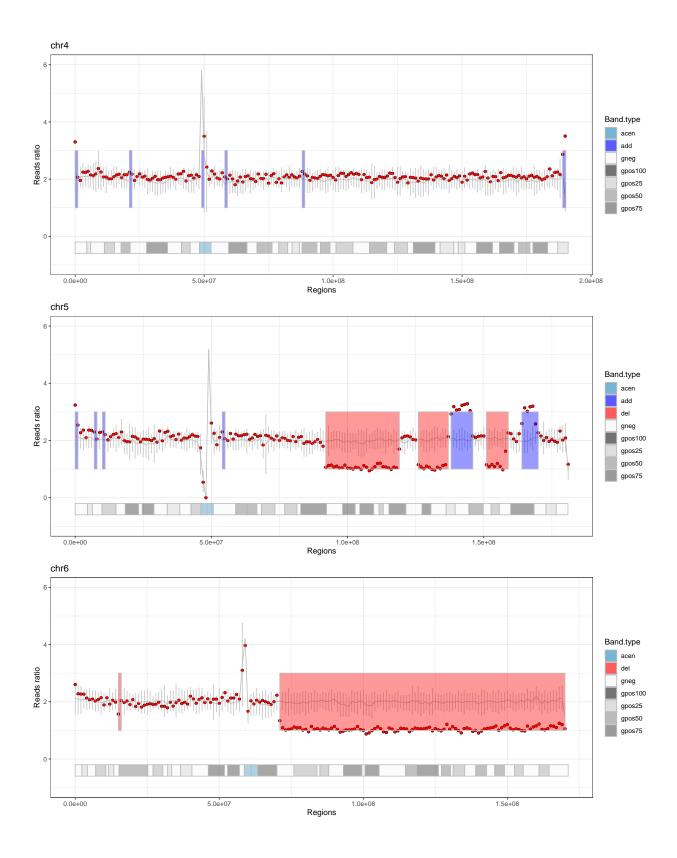
Papaemmanuil et al. (2016): Genomic Classification and Prognosis in Acute Myeloid Leukemia. The New England journal of medicine 374 (23), S. 2209–2221. DOI:10.1056/NEJMoa1516192.

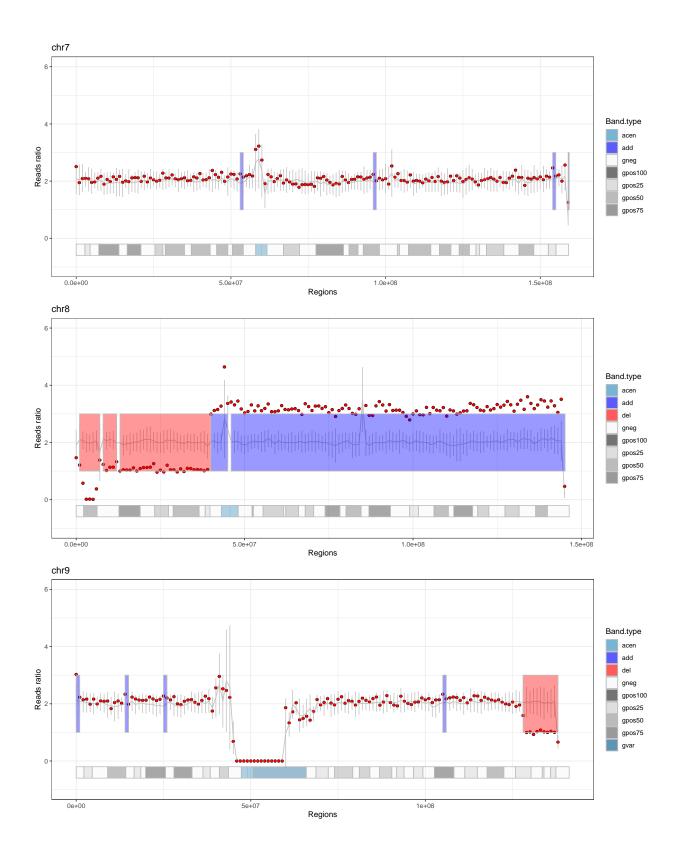
Karyotype overview:

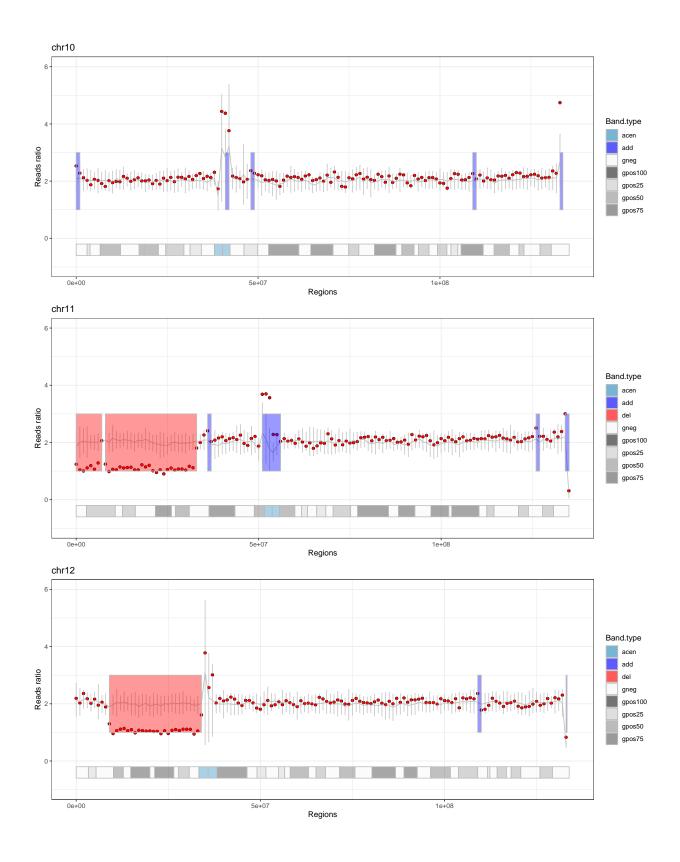


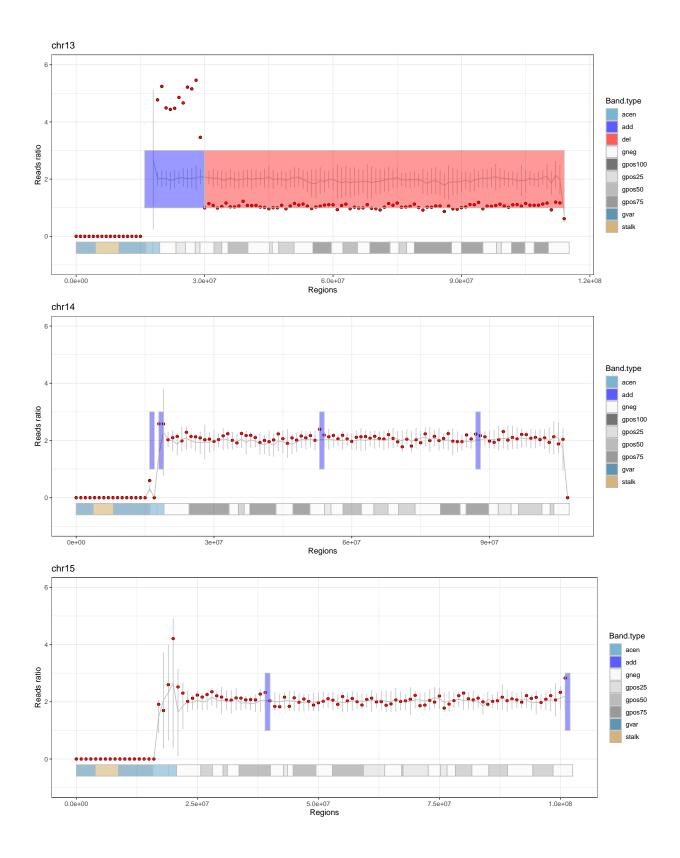
Chromosome Plots:

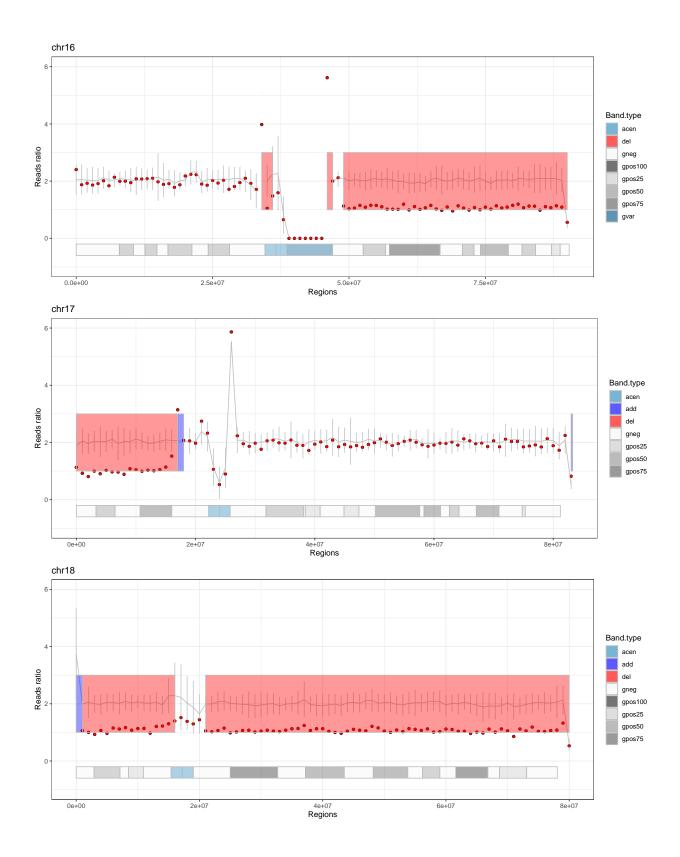


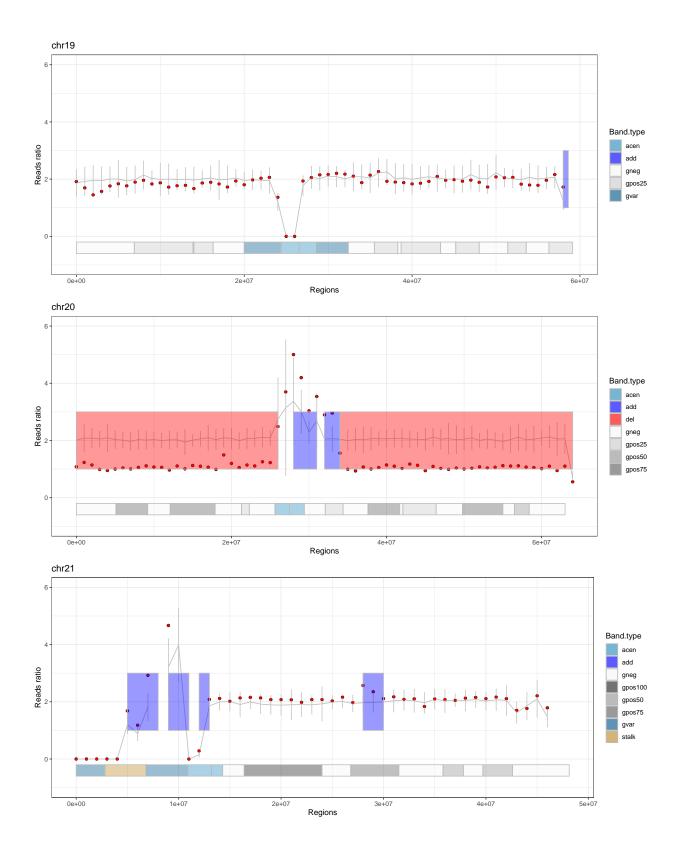


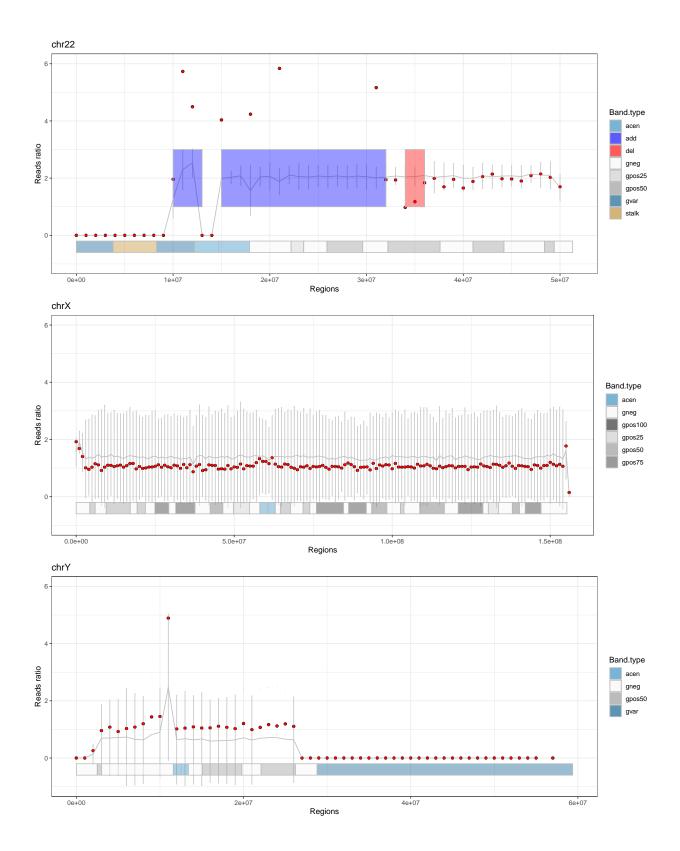












The legend contains two characteristics:

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