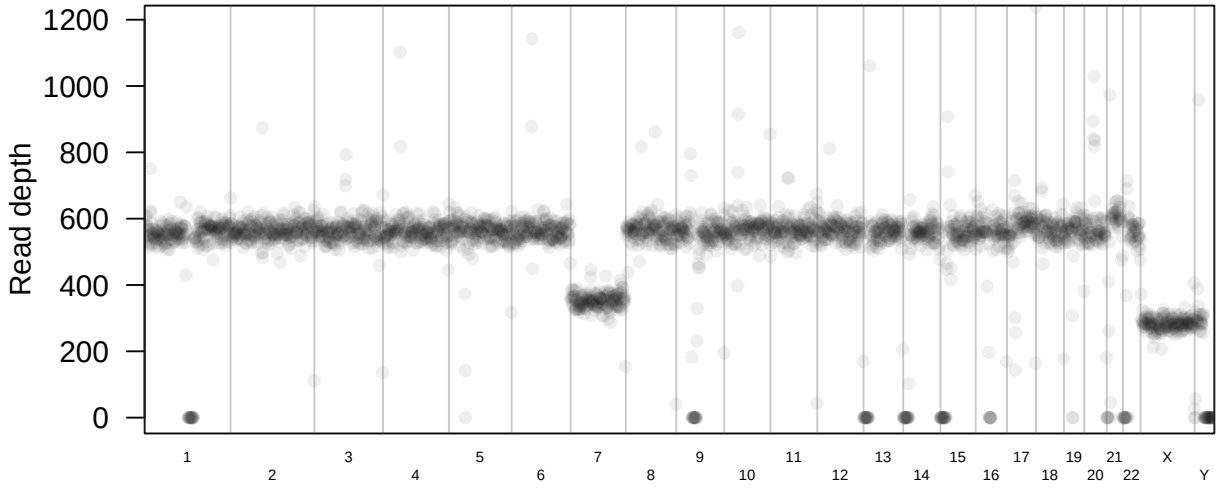
Report CORIANDR: ChrOmosomal abeRration Identifier AND Reporter in R

16-07-2023, 13:38

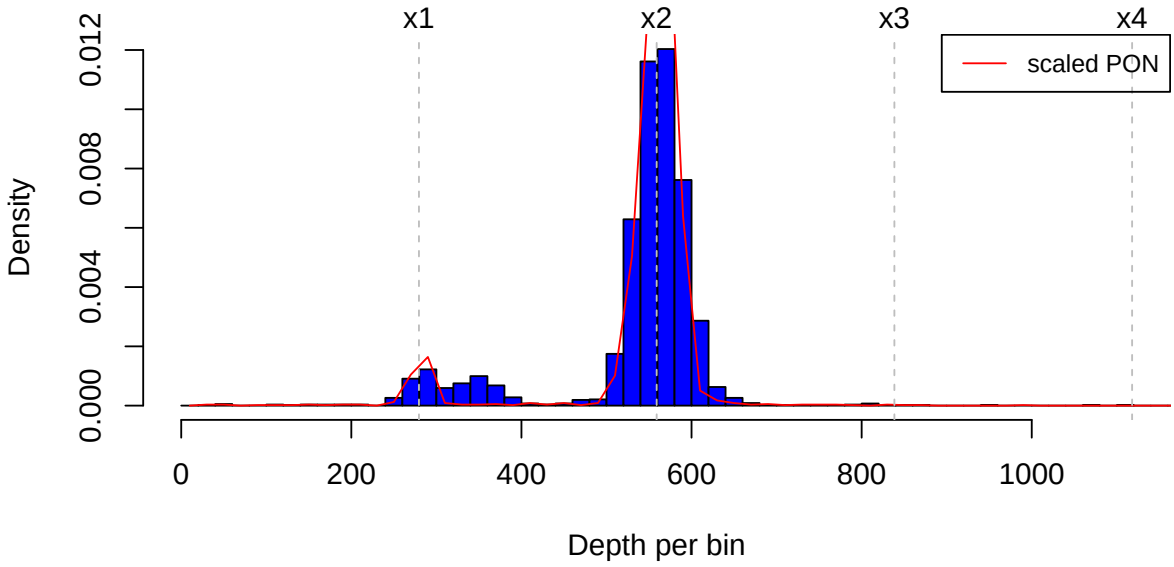
Whole genome sequencing depth of patient data



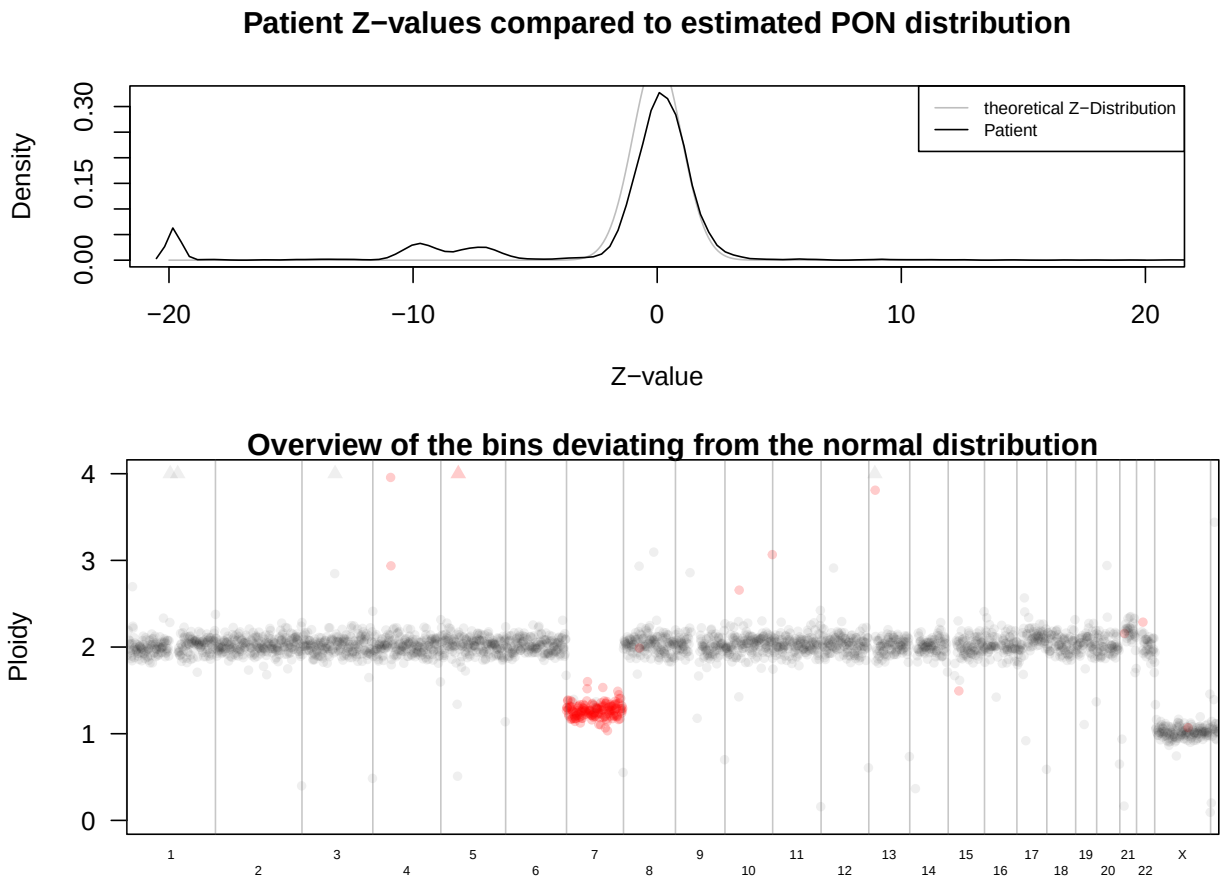
Sample sequencing characteristics and mapping statistics

Sample characterisic	Value
Sample name	**AML6**
Sample gender	**m**
Raw read pairs	1656478
Average read length	146,786
Unique mapping pairs	807.942

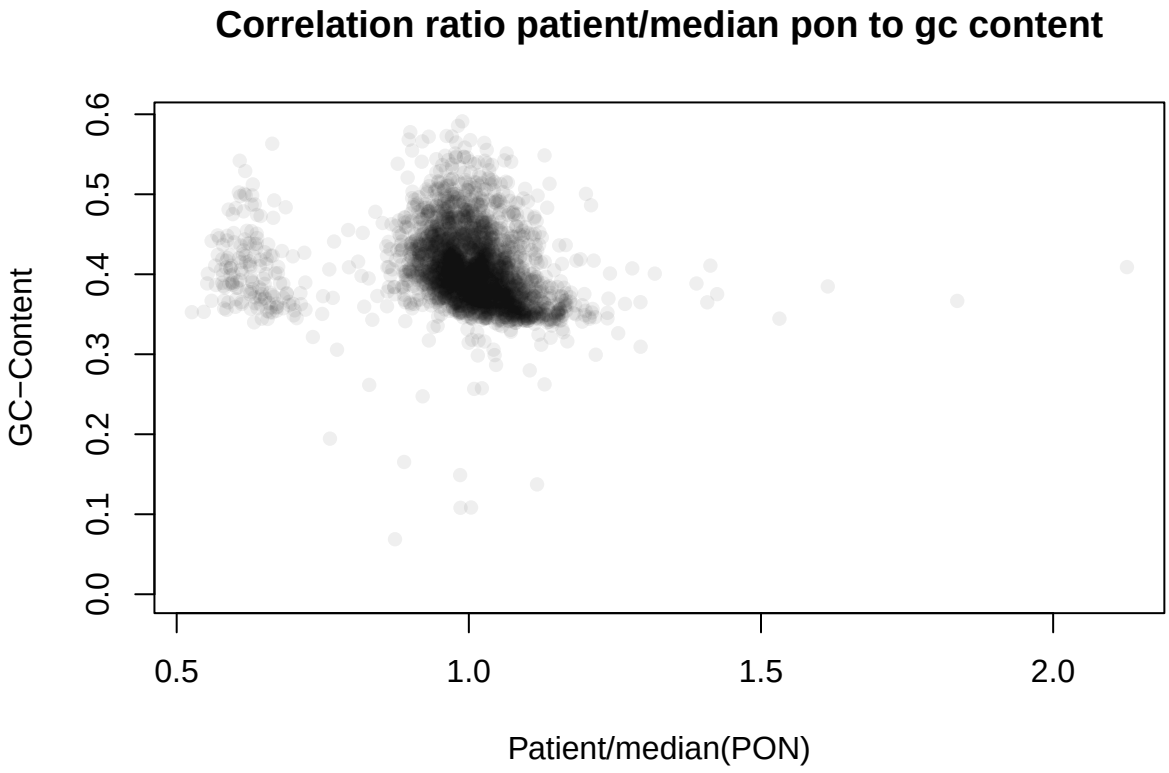
Distribution of sequencing depth per bin in sample



This graph shows the distribution of the bins according to the depth of the sequencing. The red curve represents the distribution of the PON scaled to the same depth. Ideally, the histogram and the red curve match completely. If not, this can indicate larger deletions (the first peak decreases) or larger amplifications (the third peak increases). The indications “x1, x2, x3, x4” indicate the number of copies.



In this graphic, the bins are shown in red that are outside the normal distribution after applying a Gauss test and after p-value adjustment according to the Benjamini-Hochberg method.


Correlation ratio patient/median pon to gc content
GC−Content
0.0
0.1
0.2
0.3
0.4
0.5
0.6
0.5
1.0
1.5
2.0
Patient/median(PON)

Calculated numerical karyotype:

amp(4)(q11q11)

del(7)(p22.3q36.3)

del(10)(q11.1q11.1)

− 7

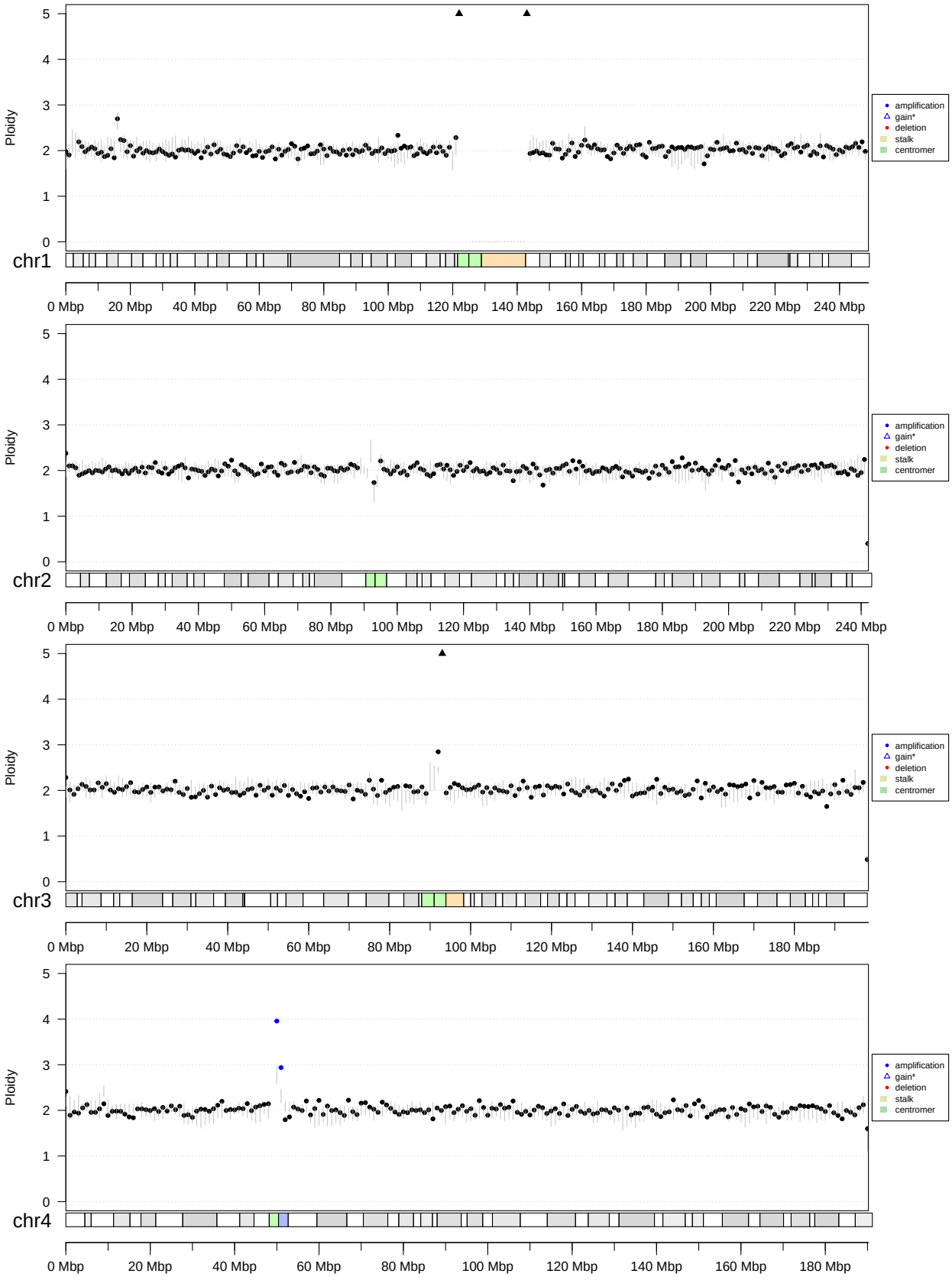
Genes affected by CNVs*:

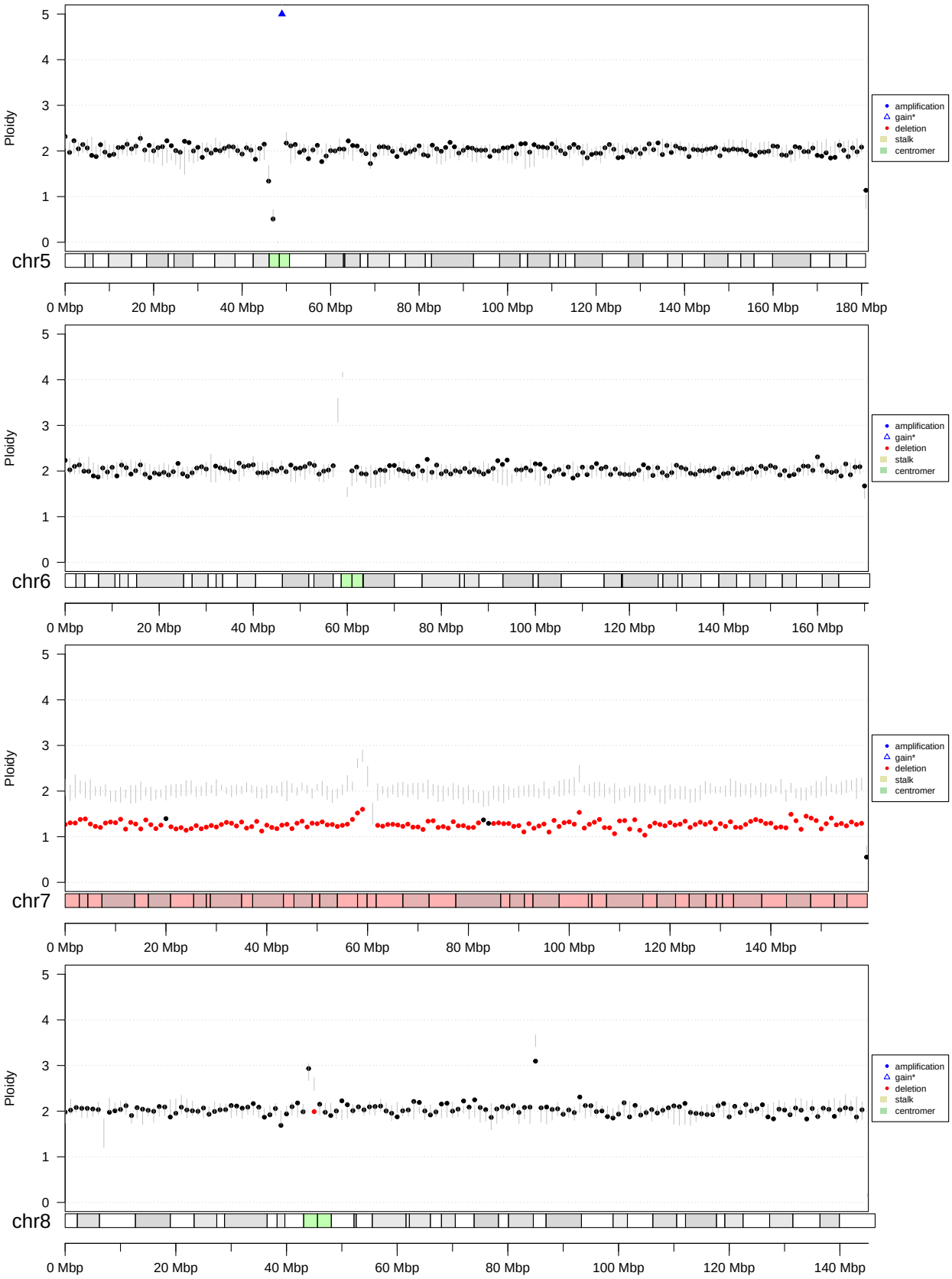
	id	symbol	chr	start	end	aberration
21587	ENSG00000164853	UNCX	chr7	1.232.872	1.237.326	del
21626	ENSG00000198286	CARD11	chr7	2.906.141	3.043.945	del
21681	ENSG00000122512	PMS2	chr7	5.970.925	6.009.106	del
21696	ENSG00000136238	RAC1	chr7	6.374.527	6.403.967	del
22481	ENSG00000146648	EGFR	chr7	55.019.017	55.211.628	del
22928	ENSG00000263001	GTF2I	chr7	74.650.231	74.760.692	del
23074	ENSG00000019991	HGF	chr7	81.699.010	81.770.438	del
23552	ENSG00000105851	PIK3CG	chr7	106.865.278	106.908.980	del
23659	ENSG00000105976	MET	chr7	116.672.196	116.798.386	del
23865	ENSG00000128602	SMO	chr7	129.188.633	129.213.545	del
24082	ENSG00000157764	BRAF	chr7	140.719.327	140.924.928	del
24217	ENSG00000197993	KEL	chr7	142.941.114	142.962.363	del
24328	ENSG00000055130	CUL1	chr7	148.697.914	148.801.110	del
24329	ENSG00000106462	EZH2	chr7	148.807.383	148.884.321	del
24436	ENSG00000106615	RHEB	chr7	151.466.012	151.520.120	del
24449	ENSG00000055609	KMT2C	chr7	152.134.922	152.436.005	del

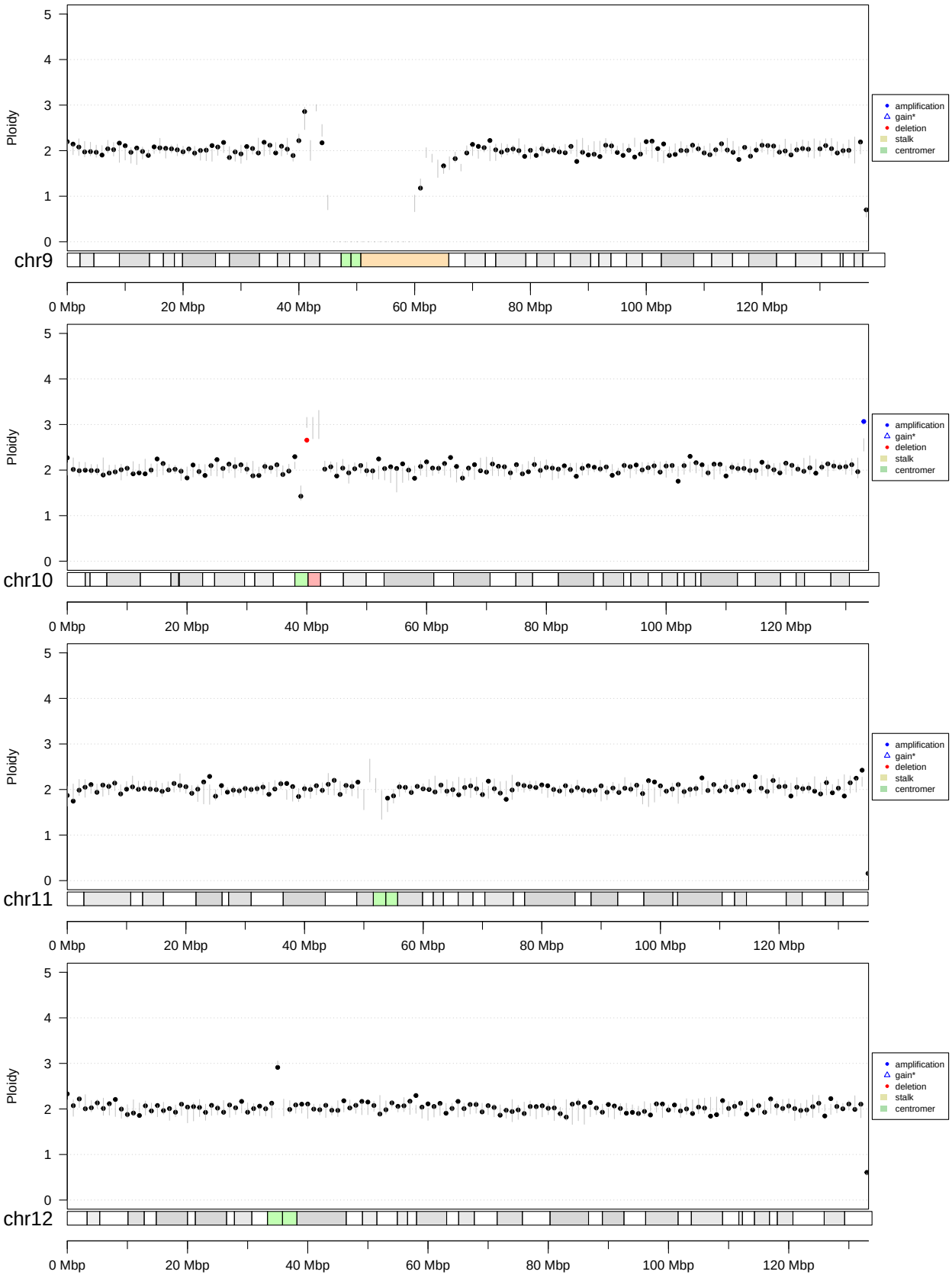
Bailey et al. (2018): Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell 173 (2), 371-385.e18. DOI: 10.1016/j.cell.2018.02.060.;

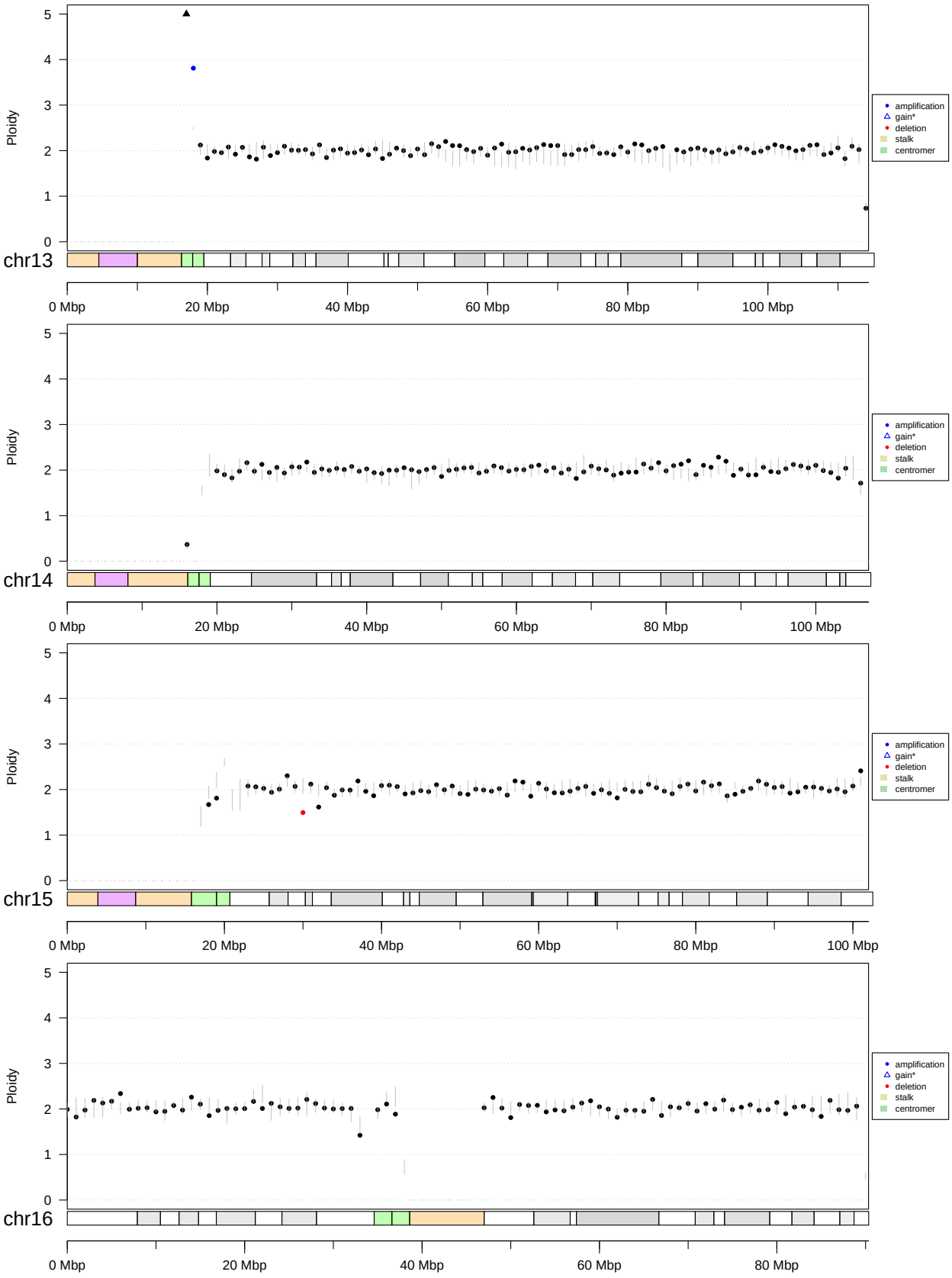
Papaemmanuil et al. (2016): Genomic Classification and Prognosis in Acute Myeloid Leukemia. The New England journal of medicine 374 (23), S. 2209–2221. DOI:10.1056/NEJMoa1516192.

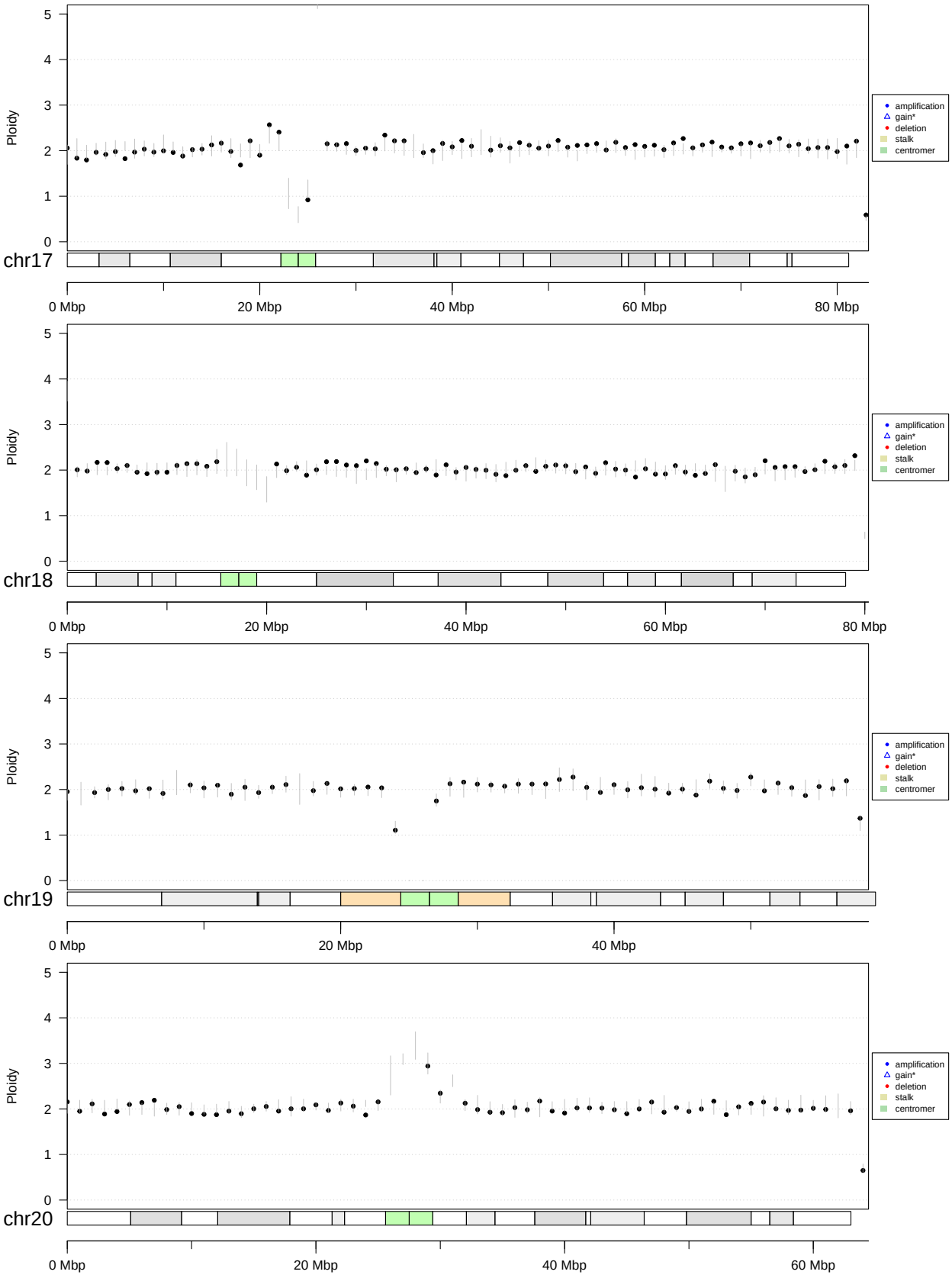
Chromosome overview plots:

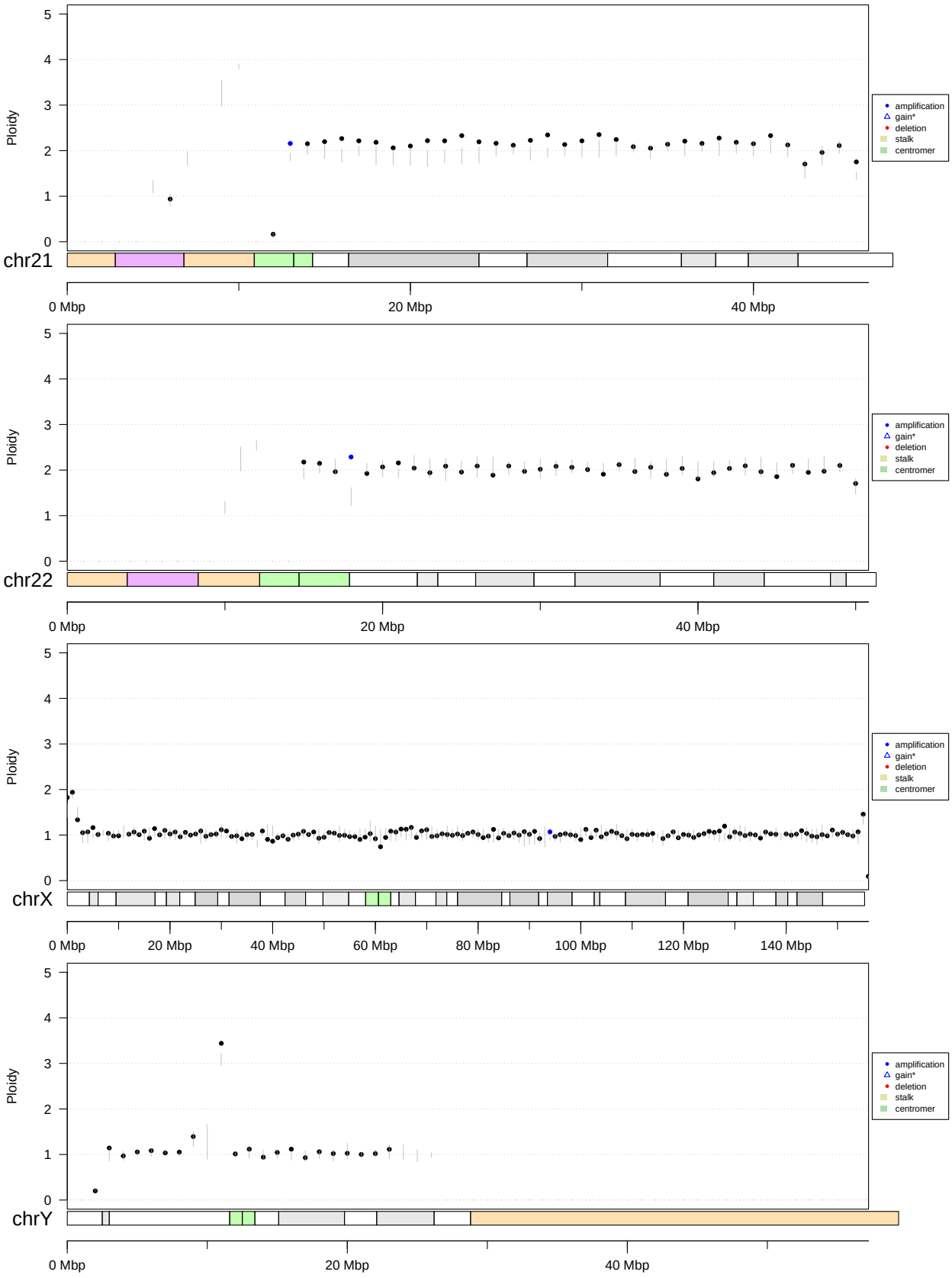



Ploidy
chr5
chr6
chr7
chr8
amplification
gain*
deletion
stalk
centromer
0 Mbp
20 Mbp
40 Mbp
60 Mbp
80 Mbp
100 Mbp
120 Mbp
140 Mbp
160 Mbp
180 Mbp


Ploidy
chr9
chr10
chr11
chr12
0 Mbp
20 Mbp
40 Mbp
60 Mbp
80 Mbp
100 Mbp
120 Mbp
amplification
gain*
deletion
stalk
centromer


Ploidy
0
1
2
3
4
5
chr13
chr14
chr15
chr16
0 Mbp
20 Mbp
40 Mbp
60 Mbp
80 Mbp
100 Mbp
amplification
gain*
deletion
stalk
centromer


Ploidy
0
1
2
3
4
5
chr17
0 Mbp
20 Mbp
40 Mbp
60 Mbp
80 Mbp
amplification
gain*
deletion
stalk
centromer
Ploidy
chr18
0 Mbp
20 Mbp
40 Mbp
60 Mbp
80 Mbp
Ploidy
chr19
0 Mbp
20 Mbp
40 Mbp
Ploidy
chr20
0 Mbp
20 Mbp
40 Mbp
60 Mbp


Ploidy
0
1
2
3
4
5
chr21
chr22
chrX
chrY
amplification
gain*
deletion
stalk
centromer
0 Mbp
20 Mbp
40 Mbp
60 Mbp
80 Mbp
100 Mbp
120 Mbp
140 Mbp

* The reads that are marked with triangles have a particularly deep sequencing depth, i.e. there are more than 4 copies.

** The legend contains characteristics to Giemsa stain results. Recognized stain values: gneg, gpos50, gpos75, gpos25, gpos100, acen, gvar, stalk.

Cheung VG, Nowak N, Jang W, Kirsch IR, Zhao S, Chen XN, Furey TS, Kim UJ, Kuo WL, Olivier M et al. Integration of cytogenetic landmarks into the draft sequence of the human genome. Nature. 2001 Feb 15;409(6822):953-8. PMID: 11237021