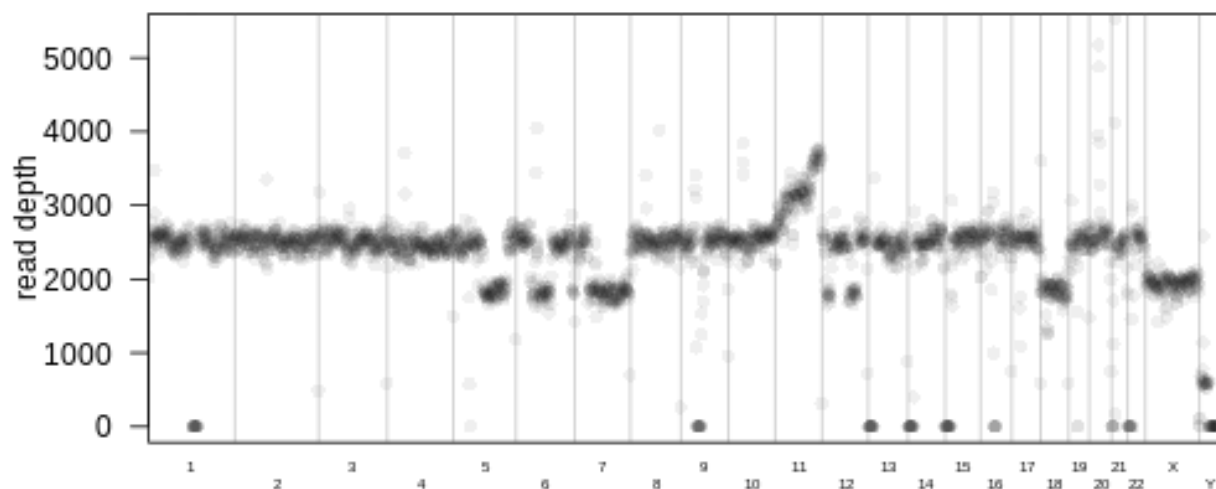# Report CORIANDR: ChrOmosomal abeRration Identifier AND Reporter in R
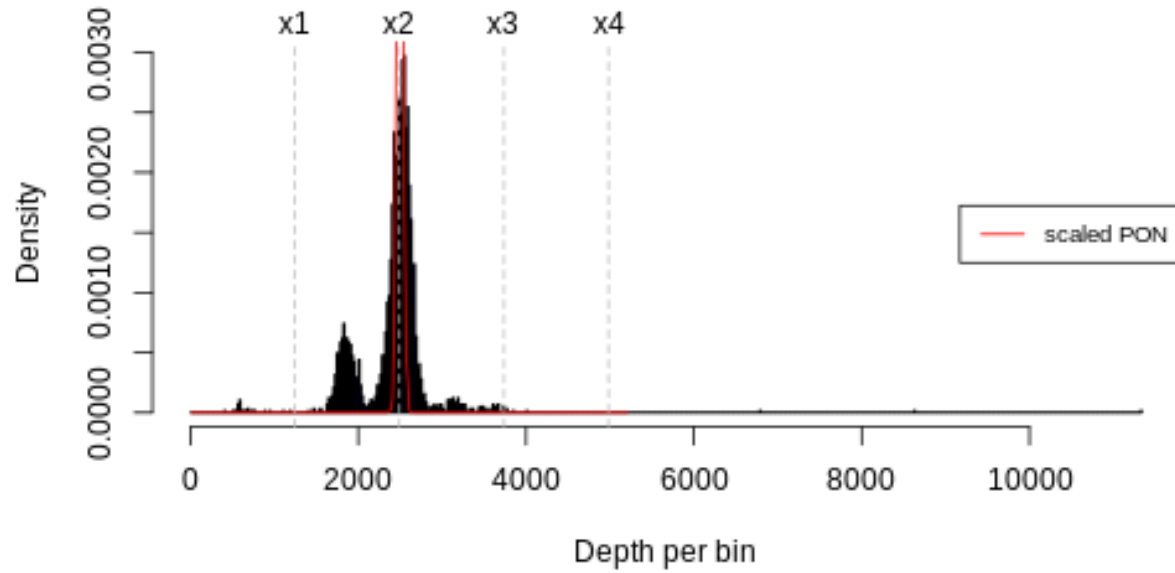
08-06-2021

Sample characteristics and mapping statistics

| Sample characterisic	Value
Sample name	**109516**
Sample gender	**m**
Raw read pairs	3.767.290
Average read length	145,304
Unique mapping pairs	3.617.856

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.

Patient z-values compared to estimated PON distribution
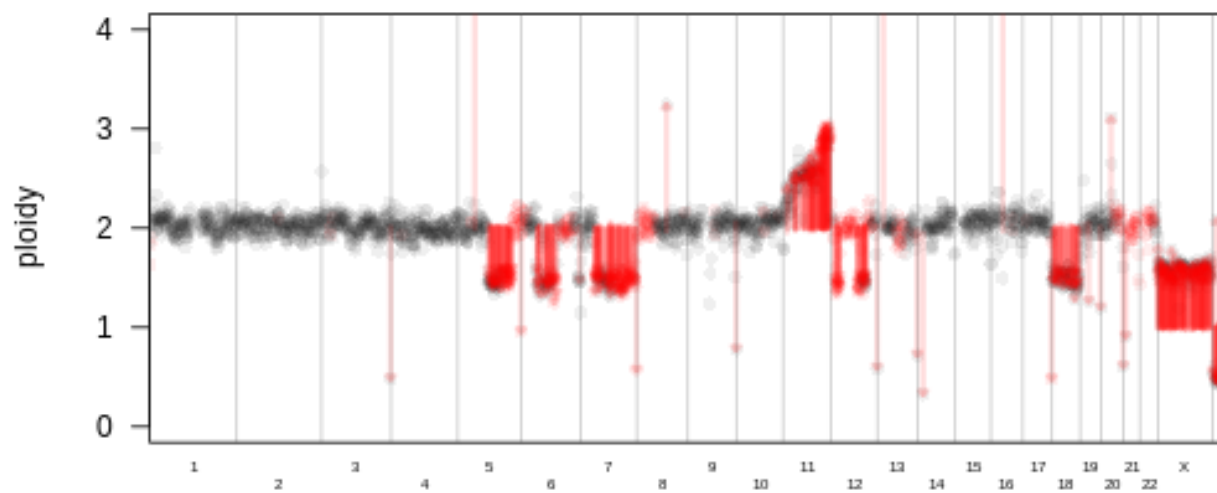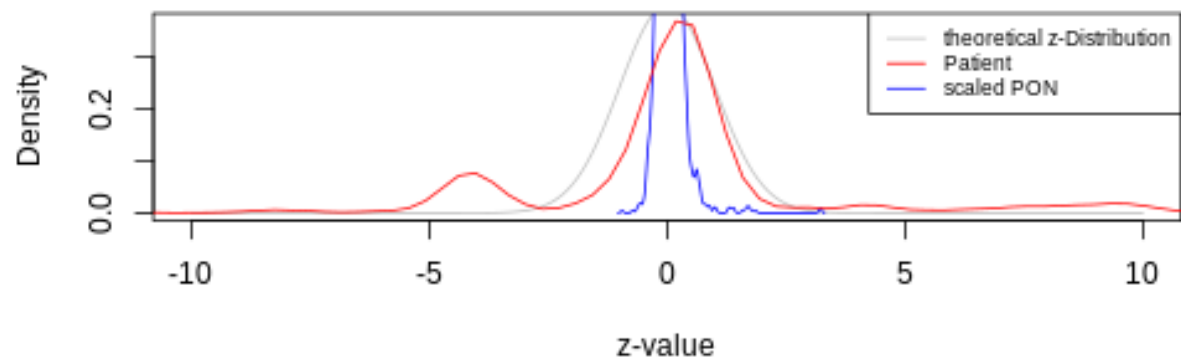
Calculated numerical karyotype:

del(1)(p36.33p36.32)

del(1)(p11.1p11.1)

add(1)(q11q11)

del(2)(q37.3q37.3)

del(3)(q12.2q12.2)

del(4)(p16.3p16.3)

del(4)(p15.32p15.32)

del(5)(p15.33p15.33)

del(5)(p15.1p15.1)

del(5)(p11p11)

del(5)(q12.2q12.2)

del(5)(q13.2q13.2)

del(5)(q14.3q33.2)

del(6)(p21.1p11.1)

del(6)(q12q16.3)

del(6)(q26q27)

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

del(7)(p14.1q36.3)

del(8)(p23.3p23.3)

del(8)(q24.3q24.3)

del(9)(q34.12q34.3)

del(10)(p15.2p15.2)

del(10)(q11.21q11.21)

del(10)(q26.3q26.3)

del(11)(p15.5p15.5)

del(12)(p13.2p11.23)

del(12)(p11.1q11)

del(12)(q21.2q23.2)

del(12)(q24.33q24.33)

del(13)(q14.12q14.12)

del(13)(q34q34)

del(14)(q13.2q13.2)

del(14)(q32.33q32.33)

del(15)(p11.1q11.1)

del(16)(p13.3p13.3)

del(16)(q11.2q11.2)

del(16)(q24.2q24.3)

del(17)(q25.2q25.2)

del(18)(p11.32q23)

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

del(19)(q13.12q13.12)

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

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

del(22)(p11.1p11.1)

del(22)(q13.32q13.33)

add(X)(p22.33q28)

add(Y)(p11.31q12)

Genes affected by CNVs*:

id	sym	bol chr		start	end abe	rration
2827	ENSG00000134250	NOTCH2	chr1	119.911.553	120.100.779	amp
9614	ENSG00000130294	KIF1A	chr2	240.713.761	240.821.036	del
12922	ENSG00000068078	FGFR3	chr4	1.793.293	1.808.872	del
15551	ENSG00000164362	TERT	chr5	1.253.147	1.295.047	del
16825	ENSG00000145715	RASA1	chr5	87.267.888	87.391.931	del
17123	ENSG00000134982	APC	chr5	112.707.498	112.846.239	del
17886	ENSG00000182578	CSF1R	chr5	150.053.291	150.113.372	del
17889	ENSG00000113721	PDGFRB	chr5	150.113.839	150.155.872	del
20143	ENSG00000156508	EEF1A1	chr6	73.515.750	73.523.797	del
21587	ENSG00000164853	UNCX	chr7	1.232.872	1.237.326	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
31674	ENSG00000148400	NOTCH1	chr9	136.494.433	136.546.048	del
31870	ENSG00000184956	MUC6	chr11	1.012.823	1.036.718	del
32366	ENSG00000133818	RRAS2	chr11	14.277.922	14.364.506	amp
32621	ENSG00000184937	WT1	chr11	32.387.775	32.435.885	amp
33142	ENSG00000198561	CTNND1	chr11	57.753.243	57.819.546	amp
33487	ENSG00000168066	SF1	chr11	64.764.606	64.778.786	amp
33490	ENSG00000133895	MEN1	chr11	64.803.510	64.811.294	amp
33764	ENSG00000110092	CCND1	chr11	69.641.087	69.654.474	amp
33855	ENSG00000165458	INPPL1	chr11	72.223.701	72.239.147	amp
34405	ENSG00000082175	PGR	chr11	101.029.624	101.130.524	amp
34521	ENSG00000149311	ATM	chr11	108.222.484	108.369.102	amp
34754	ENSG00000118058	KMT2A	chr11	118.436.464	118.526.832	amp
37950	ENSG00000111276	CDKN1B	chr12	12.715.058	12.722.369	del
37989	ENSG00000171681	ATF7IP	chr12	14.365.632	14.502.935	del
38141	ENSG00000133703	KRAS	chr12	25.204.789	25.250.936	del
39959	ENSG00000179295	PTPN11	chr12	112.418.351	112.509.913	amp
40502	ENSG00000177084	POLE	chr12	132.623.753	132.687.365	del
43977	ENSG00000142208	AKT1	chr14	104.769.349	104.795.751	del
46467	ENSG00000103126	AXIN1	chr16	287.440	352.723	del
46622	ENSG00000103197	TSC2	chr16	2.047.967	2.089.491	del
52042	ENSG00000141579	ZNF750	chr17	82.829.434	82.840.022	del
52137	ENSG00000177426	TGIF1	chr18	3.411.608	3.459.978	del
52741	ENSG00000152217	SETBP1	chr18	44.680.173	45.068.510	del
52799	ENSG00000175387	SMAD2	chr18	47.808.957	47.931.146	del
52876	ENSG00000141646	SMAD4	chr18	51.028.394	51.085.045	del
53051	ENSG00000171791	BCL2	chr18	63.123.346	63.320.128	del
54791	ENSG00000099821	POLRMT	chr19	617.221	633.537	del
54831	ENSG00000118046	STK11	chr19	1.177.558	1.228.435	del

id	sym	bol chr		start	end abe	rration
54850	ENSG00000071626	DAZAP1	chr19	1.407.569	1.435.687	del
54949	ENSG00000088256	GNA11	chr19	3.094.362	3.123.999	del
54995	ENSG00000167658	EEF2	chr19	3.976.056	3.985.463	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 Plots:

chr1

chr2

chr3

chr4

chr5

chr16

chr17

chr18

chr19

chr20

11

chr21

chr22

chrX

chrY