

Generating realistic data with known truth using the `jointseg` package

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September 9, 2014

Abstract

This vignette illustrates how the `jointseg` package may be used to generate a variety of copy-number profiles from the same biological “truth”. Such profiles have been used to compare the performance of segmentation methods [1].

```
library(jointseg)
```

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1 Setup

The parameters are defined as follows:

```
n <- 1e4                                ## signal length
bkp <- c(2334, 6121)                      ## breakpoint positions
regions <- c("(1,1)", "(1,2)", "(0,2)")    ## copy number regions

ylims <- cbind(c(0, 5), c(-0.1, 1.1))
colG <- rep("#88888855", n)
hetCol <- "#00000088"
```

For convenience we define a custom plot function for this vignette:

```
plotFUN <- function(dataSet, tumorFraction) {
  regDat <- loadCnRegionData(dataSet=dataSet, tumorFraction=tumorFraction)
  sim <- getCopyNumberDataByResampling(n, bkp=bkp, regions=regions, regData=regDat)
  dat <- sim$profile
  wHет <- which(dat$genotype==1/2)
  colGG <- colG
  colGG[wHет] <- hetCol
  plotSeg(dat, sim$bkp, ylims=ylims, col=colGG)
}
```

```
tf <- 1  
plotFUN(ds, tf)
```

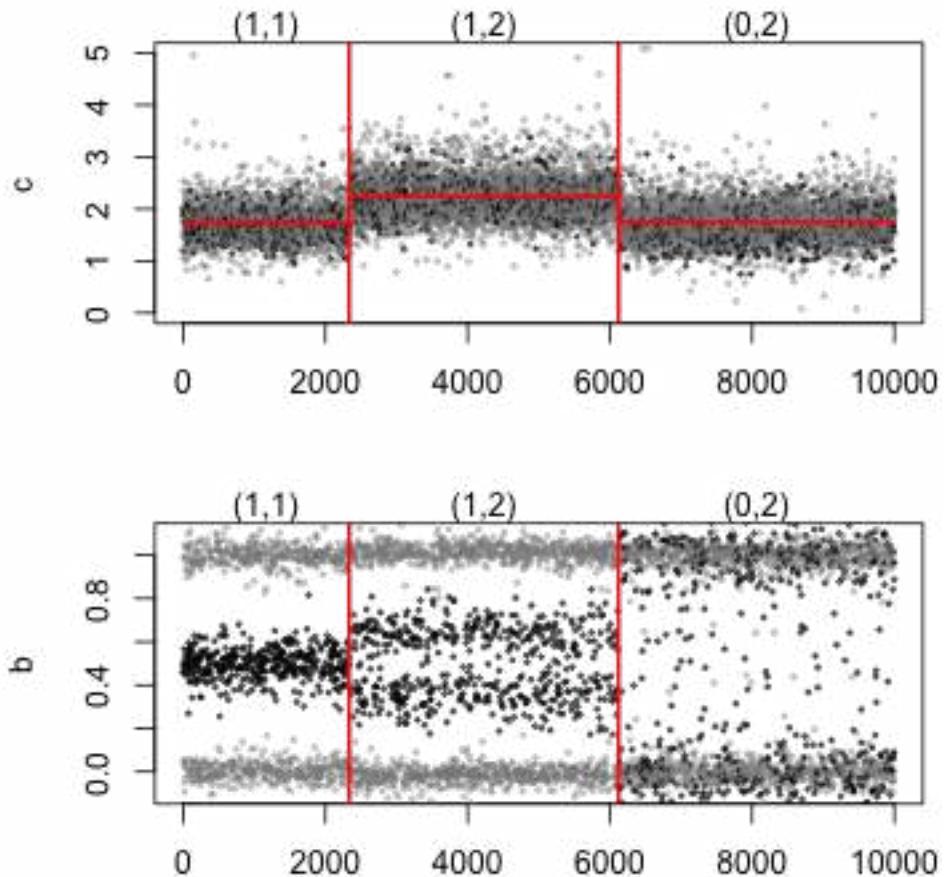


Figure 1: Data set GSE29172, 100% tumor cells

2 Affymetrix data

```
ds <- "GSE29172"
```

```
tf <- 1  
plotFUN(ds, tf)
```

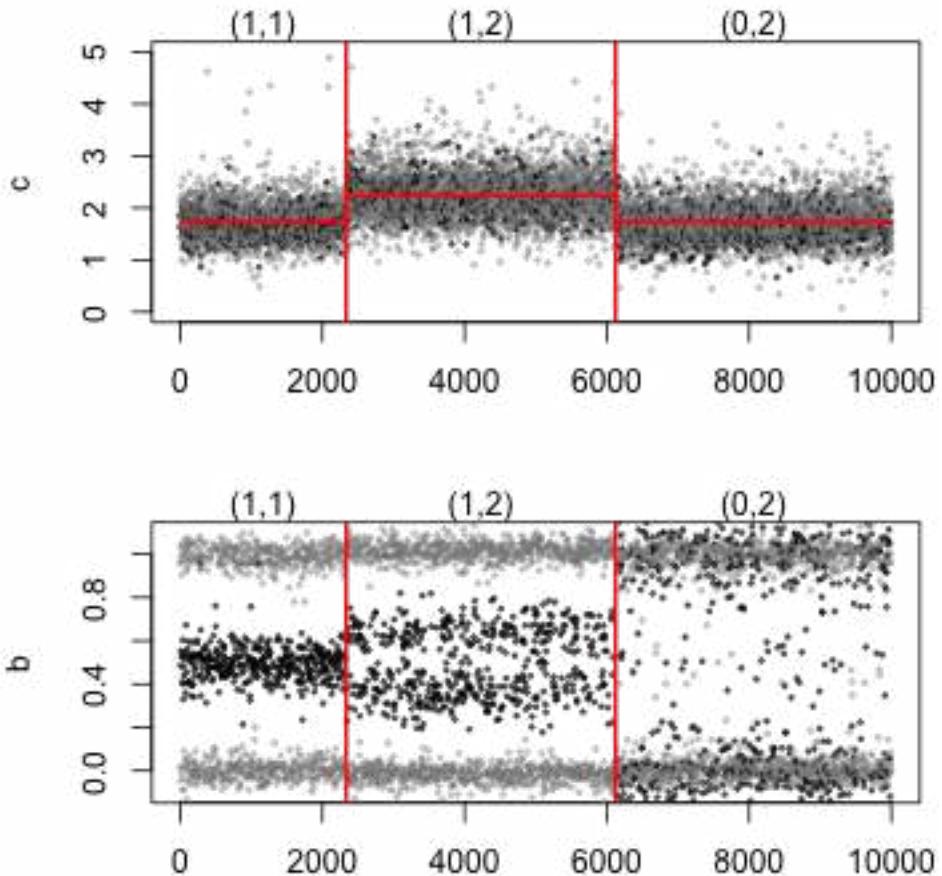


Figure 2: Data set GSE29172, 100% tumor cells (another resampling)

```
tf <- .7  
plotFUN(ds, tf)
```

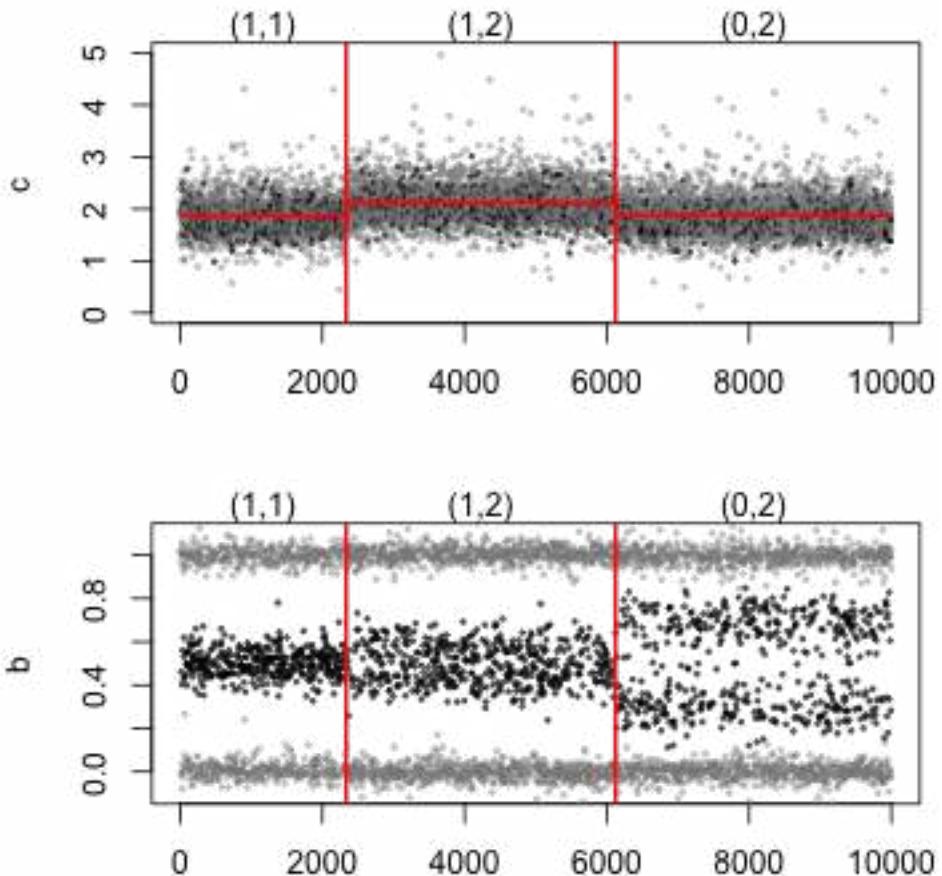


Figure 3: Data set GSE29172, 70% tumor cells

```
tf <- .5  
plotFUN(ds, tf)
```

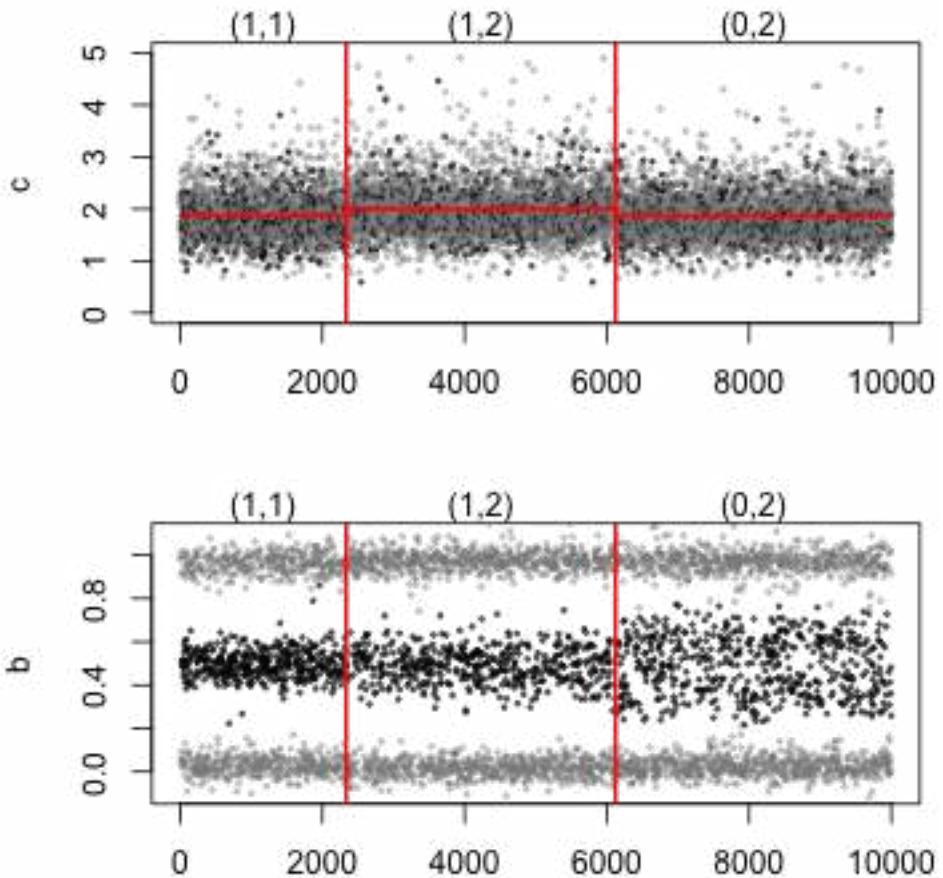


Figure 4: Data set GSE29172, 50% tumor cells

```
tf <- 1  
plotFUN(ds, tf)
```

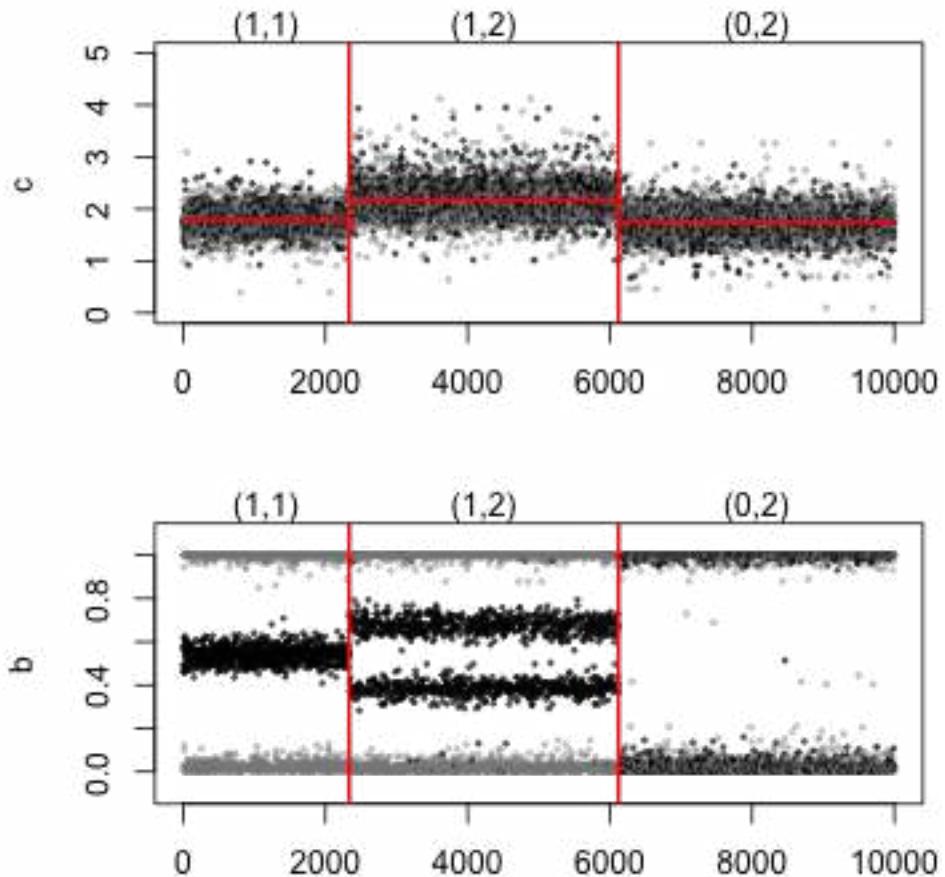


Figure 5: Data set GSE11976, 100% tumor cells

3 Illumina data

```
ds <- "GSE11976"
```

References

- [1] Morgane Pierre-Jean, Guillem J Rigaill, and Pierre Neuvial. Performance evaluation of DNA copy number segmentation methods. *Briefings in Bioinformatics*, to appear.

A Session information

```
sessionInfo()  
## R version 3.1.1 (2014-07-10)
```

```
tf <- 1  
plotFUN(ds, tf)
```

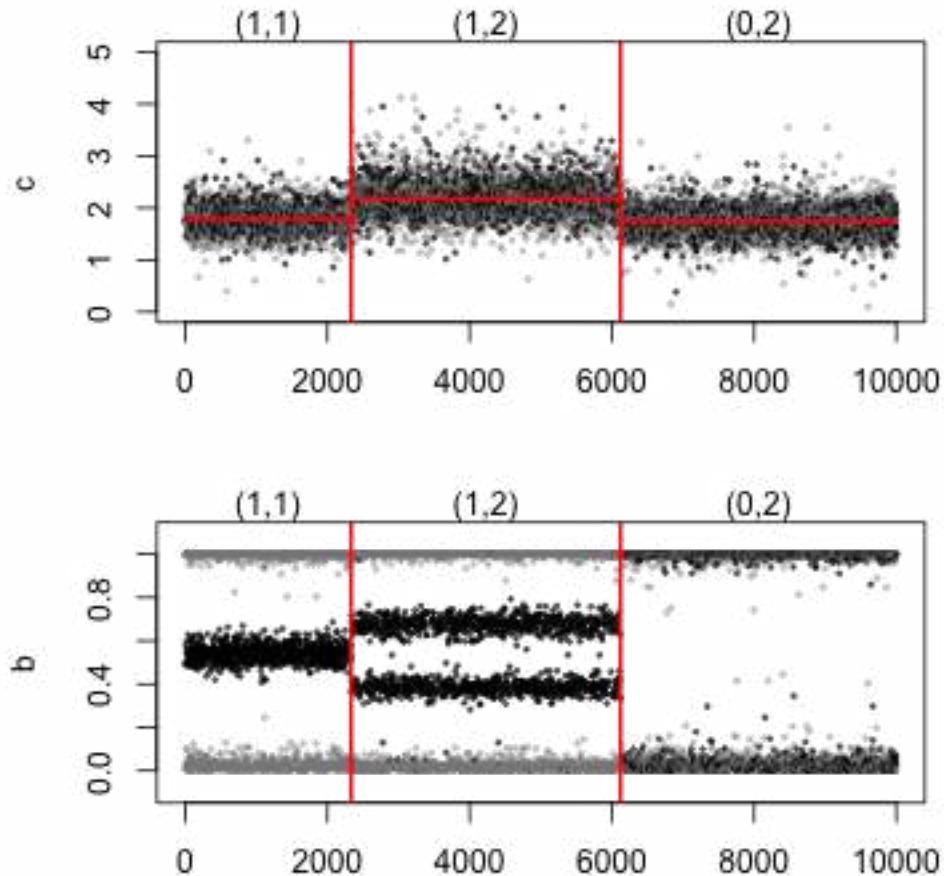


Figure 6: Data set GSE11976, 100% tumor cells (another resampling)

```
tf <- .79  
plotFUN(ds, tf)
```

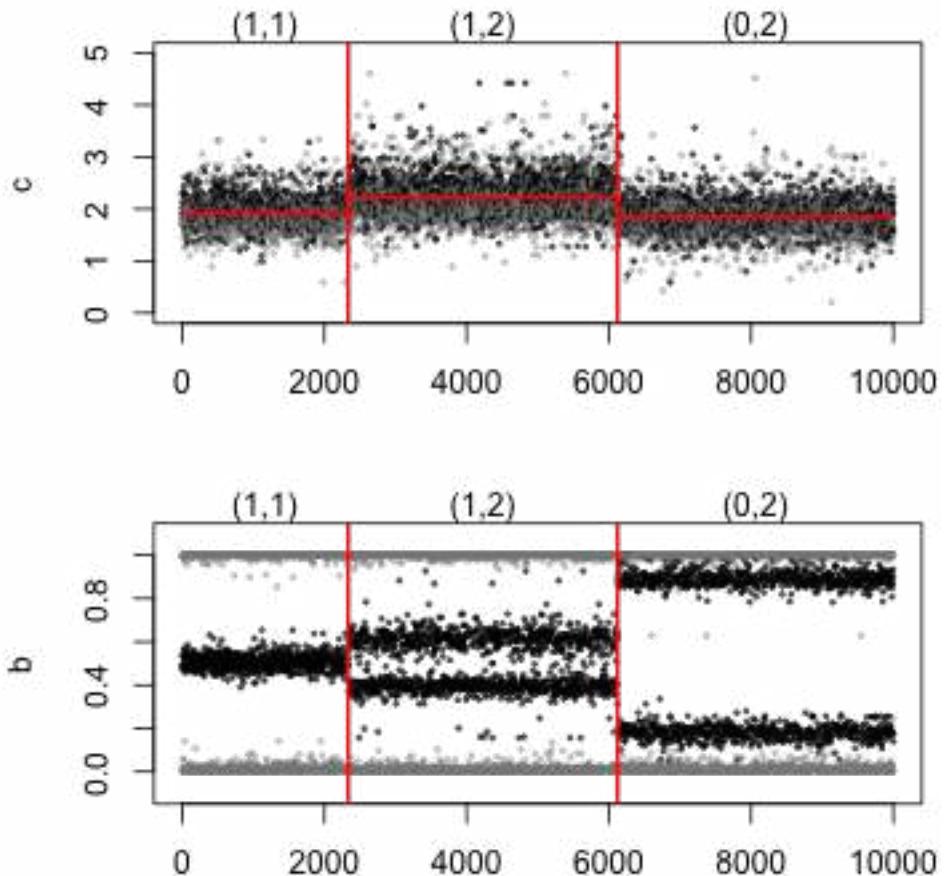


Figure 7: Data set GSE11976, 79% tumor cells

```
tf <- .5  
plotFUN(ds, tf)
```

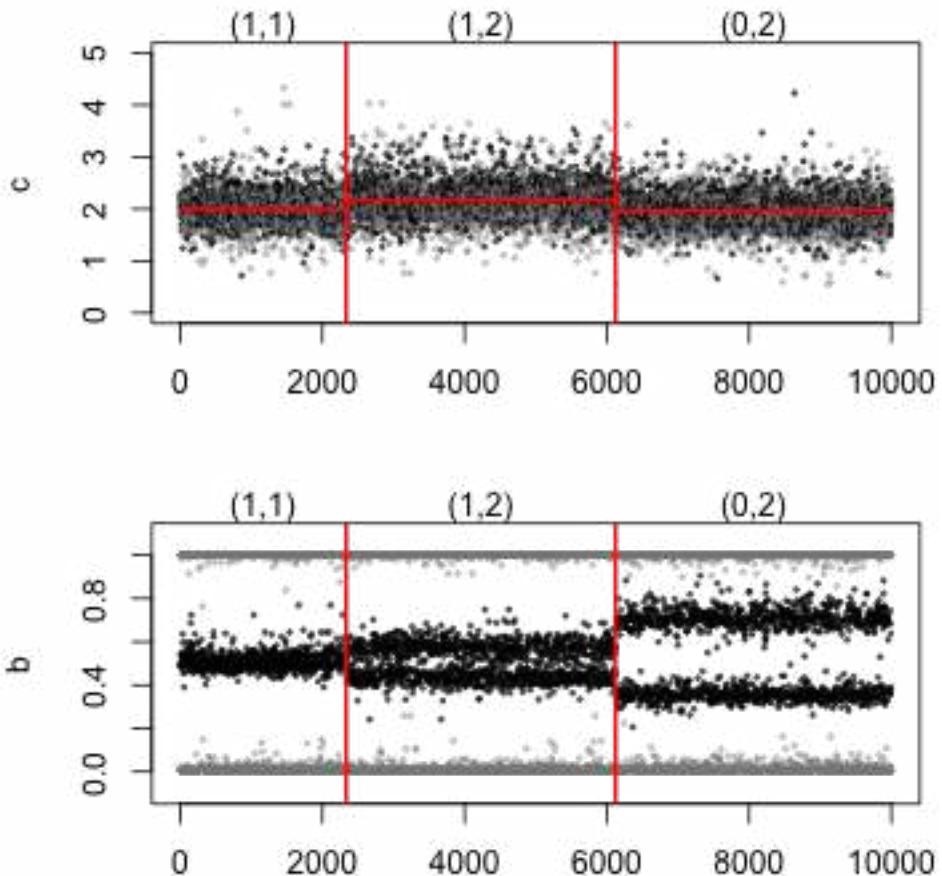


Figure 8: Data set GSE11976, 50% tumor cells

```
## Platform: x86_64-apple-darwin10.8.0 (64-bit)
##
## locale:
## [1] fr_FR.UTF-8/fr_FR.UTF-8/fr_FR.UTF-8/C/fr_FR.UTF-8/fr_FR.UTF-8
##
## attached base packages:
## [1] stats      graphics   grDevices utils      datasets   base
##
## other attached packages:
## [1] jointseg_0.6.1    acnr_0.2.0       R.utils_1.33.0    R.oo_1.18.2
## [5] R.methodsS3_1.6.2 knitr_1.6.10
##
## loaded via a namespace (and not attached):
## [1] DNAcopy_1.38.1    evaluate_0.5.5    formatR_1.0       highr_0.3.1
## [5] matrixStats_0.10.1 methods_3.1.1     stringr_0.6.2     tools_3.1.1
```