

Estimate contrast with known alpha

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Goal of the study

In this report, our goal is to check the distribution of phylogenetic contrasts with known alpha, known shift configuration and unknown shift values that are used in the bootstrap procedure in the `l1ou` package. In the last report, we used the true shift edge, true shift values, true alpha.

We used code from the bootstrap procedure to simulate contrast values. Then we calculated the mean and standard deviation of the contrast values at each node. The conclusions above held for all nodes, whether nodes had a shift on a child edge or not.

Estimate shift configuration from original lizard data (change)

We used the lizard tree from the `phylolm` package and associated trait data on these lizard species (just the first trait, which is the first PC axis from a PCA). To simulate data according to a model that is biologically plausible, we estimated the shift configuration, shift values and covariance parameters from original lizard data.

This trait was analyzed to estimate the shifts in trait evolution using the function `fit_OU` from the `l1ou` package.

```
sessionInfo()$otherPkgs$l1ou$Version #version 1.29
```

```
## [1] "1.29"
```

```
load("eModel_2_27.RData") # eModel.tree same as lizard.tree
```

```
data(lizard.tree, lizard.traits)
```

```
lizard <- adjust_data(lizard.tree, lizard.traits[,1])
```

```
## the new tree is normalized: each tip at distance 1 from the root.
```

```
## new Y: matrix of size 100 x 1
```

```
n_tips=length(lizard$tree$tip.label) # 100: total number of tips
```

```
truealpha=eModel$alpha # 0.606897967627925
```

```
y0=eModel$intercept
```

```
truetheta = y0 + l1ou::convert_shifts2regions(eModel$tree,  
                                              eModel$shift.configuration, eModel$shift.values)
```

```
table(truetheta)
```

```
## truetheta
```

```
## -5.25186223573345 -4.56654712450638 -4.04766913950491 -3.45016309674712
```

```
## 1 3 1 1
```

```
## -3.23947780027265 -2.72293353502748 -2.36362306302032 -1.6605679645554
```

```
## 1 7 1 25
```

```
## 0.248809571469948
```

```
## 158
```

```
# -5.25186223573345 -4.56654712450638 -4.04766913950491 -3.45016309674712 -3.23947780027265 -2.72293353502748  
# 1 3 1 1 1
```

```

#-2.36362306302032  -1.6605679645554  0.248809571469948
#                      1                      25                      158
nShifts    = length(eModel$shift.configuration) # c(55,98,118,74,14,77,32,164)

sigma2=eModel$sigma2    # 0.0625186613743141
shift_config=eModel$shift.configuration
shiftnode=eModel$tree$edge[shift_config,1]# internal nodes with shift

```

Below, we use this model to simulate new data using the `rTraitCont` function from the `phylolm` package. RE is the result of the function `sqrt_OU_covariance`, which calculates the square-root of the phylogenetic covariance matrix with a recursive algorithm, which traverses the tree once. `covInverseSqrt` is the inverse square root of the phylogenetic covariance matrix, and `covSqrt` is the square-root of the phylogenetic covariance matrix. Finally, `contrast` contains the contrasts at all nodes. These matrices and contrasts were obtained using the true value of α , the same value used to simulate the data. This is an ideal situation when α is known without error.

simulation procedure

We use the same data for all scenarios: `set.seed` is used here to make it possible to re-simulate the exact same data, then these data are saved in a file (`Y_table`).

```

n_sim=100000
Y_table=matrix(nrow=n_sim, ncol=n_tips, data=NA)
set.seed(1293)
for (i in 1:n_sim) {
  Y <- rTraitCont(eModel$tree, "OU", theta=truetheta,
                  alpha=truealpha,
                  sigma=sqrt(eModel$sigma2), root.value=y0)
  Y_table[i,]=Y
}
colnames(Y_table)=eModel$tree$tip.label
save(Y_table, file="Y_table_2_27.RData")

load("Y_table_2_27.RData")
len=dim(Y_table)[1]
n_sim=100000

```

Just to check our simulated values, we define our own function “ss” to calculate the variance of assuming a known population mean of 0. The name “ss” stands for “sum of squares”, although this sum of squares is rescaled by the sample size.

```

ss=function(x){
  sum(x^2)/length(x)
}

```

Now to check the mean and variance of the simulated Y values:

```

round(colMeans(Y_table-rep(eModel$mu[,1],each=n_sim),5)) # all 0: good
convar = apply(Y_table-rep(eModel$mu[,1],each=n_sim), 2, ss)
round(convar,3) # all about the same
mean(convar) # 0.03622244: same as expected variance below: good
sigma2/(2*truealpha) * (1-exp(-2*truealpha*1)) # 0.03620576

```

Contrast calculation procedure

We consider 4 scenarios for the calculation of contrasts, depending on what parameters are known (yes) or unknown (no) for the calculation of the inverse square-root of the covariance matrix.

	1: full model known	2: alpha known	3: shift config known	4: tree known
β_0, β_1, \dots	yes	no	no	no
α	yes	yes	no	no
shift config	yes	yes	yes	no

For the tree topology, we assume it known always. Refer to “filename” for the results of simulation 1, in which we know everything, and the contrasts behave as expected.

scenario 1: all parameters known and set to their true values

use Y table, call contrasts table “contrast_table1”

```
REf = sqrt_OU_covariance(lizard.tree, alpha=truealpha,
                          root.model = "OUfixedRoot", # eModel$l1ou.options$root.model,
                          check.order=F, check.ultrametric=F) # needed in l1ou v1.29

covInverseSqrt = t(REf$sqrtInvSigma)
```

To check the correctness of this square-root covariance matrix calculation:

```
treeheight = branching.times(eModel$tree)[1] # 1.00000000002
Vbm = vcv(eModel$tree) # t_ij values. Tree height is 1 here, do d_ij = 2*(1-t_ij)
Vou = (1-exp(-2*truealpha*Vbm)) * exp(-truealpha*2*(treeheight-Vbm)) / (2*truealpha)
tmp = covInverseSqrt %*% Vou %*% t(covInverseSqrt)
diag(tmp) # if normalize.tree.height=T was omitted above, and l1ou v1.29:
# all 1.742289: ratio found later in convar/sigma2
all.equal(tmp, diag(n_tips), scale=1) # TRUE
```

Now that we can trust the inverse square-root of the covariance matrix, 158 we use it to calculate contrasts:

```
contrast_table=matrix(nrow=n_sim, ncol=n_tips, data=NA)
for (i in 1:len) {
  contrast_table[i,]=covInverseSqrt%*%(Y_table[i,] - eModel$mu)
}
save(contrast_table, file="contrast_table_2_27.RData")
```

scenario 2: beta unknown

Here, we assume that we know alpha, we know the shift configuration, but beta is unknown. 171 It took around 3 hours.

```
RE = sqrt_OU_covariance(lizard$tree, alpha=truealpha, # assumed known here
                        root.model = eModel$l1ou.options$root.model,
                        check.order=F, check.ultrametric=F)
covInverseSqrt <- t(RE$sqrtInvSigma)
```

```

contrast_table2=matrix(nrow=len, ncol=n_tips, data=NA)
sigma_table2=rep(NA,len)
mu_table2=matrix(nrow=len, ncol=n_tips, data=NA)
shift_values2=matrix(nrow=len, ncol=length(shift_config), data=NA)
for (i in 1:len) {
  model= fit_OU(lizard$tree, Y_table[i,], shift_config,
               alpha.upper=truealpha, alpha.lower=truealpha, alpha.starting.value=truealpha)
  # the line above is to fix alpha to the known value
  sigma_table2[i] = model$sigma2
  shift_values2[i,]=model$shift.values
  mu_table2[i,]=model$mu
  contrast_table2[i,] <- covInverseSqrt%*%(Y_table[i,] - model$mu)
}
save(sigma_table2,file="knownalpha_sigma2.RData")
save(contrast_table2,file="knownalpha_contrast.RData")
save(mu_table2,file="knownalpha_mu.RData")
save(shift_values2,file="knownalpha_shiftvalues.RData")

```

scenario 3: alpha and beta unknown

Here we assume that the shift configuration is known, but not alpha and not beta. The correlation matrix among tips is unknown, then, so its inverse square-root needs to be re-calculated (based on the estimated alpha) for each simulated vector.

```

contrast_table3 = matrix(nrow=len, ncol=n_tips, data=NA)
alpha_table3 = rep(NA,len)
sigma_table3 = rep(NA,len)
mu_table3 = matrix(nrow=len, ncol=n_tips, data=NA)
shift_values3 = matrix(nrow=len, ncol=length(shift_config), data=NA)
# also save sigma2 estimated values, and 8 shift values? and/or 100 mu values?
for (i in 1:len) {
  model= fit_OU(lizard$tree, Y_table[i,], shift_config)
  alpha_table3[i] = model$alpha
  sigma_table3[i] = model$sigma2
  shift_values3[i,]=model$shift.values
  mu_table3[i,]=model$mu
  RE = sqrt_OU_covariance(lizard$tree, alpha=model$alpha, # alpha estimated here
                          root.model = eModel$l1ou.options$root.model,
                          check.order=F, check.ultrametric=F)
  covInverseSqrt <- t(RE$sqrtInvSigma)
  contrast_table3[i,] <- covInverseSqrt%*%(Y_table[i,] - model$mu)
}
save(contrast_table3,file="unknownalpha_contrast.RData")
save(alpha_table3, file="unknownalpha_alpha.RData")
save(sigma_table3,file="unknownalpha_sigma2.RData")
save(mu_table3,file="unknownalpha_mu.RData")
save(shift_values3,file="unknownalpha_shiftvalues.RData")

```

scenario 4.1: unknown shift configuration

Also: unknown alpha and unknown beta. The estimation of the shift configuration takes a long time, so the number of simulations had to be reduced. In a preliminary run, the contrasts for 1262 simulations were calculated in 24 hours. So for the final run, we plan to complete a total of 2000 simulations ("n_sim" below). criterion: pBIC, which is the default of function `estimate_shift_configuration`.

```
n_sim=800 # lower because estimation is a lot slower when we have to search for the config
# use first n_sim rows of Ytable only
Y_table=Y_table[1:n_sim,]
contrast_table4=matrix(nrow=n_sim, ncol=n_tips, data=NA)
vectorOfShift <- vector(mode = "list", length = n_sim)
alpha_table4 = rep(NA,n_sim)
sigma_table4 = rep(NA,n_sim)
mu_table4 = matrix(nrow=n_sim, ncol=n_tips, data=NA)
shift_values4 = vector(mode = "list", length = n_sim)

for (i in 1:n_sim) {
  model= estimate_shift_configuration(lizard$tree, Y_table[i,])
  vectorOfShift[[i]] =model$shift.configuration
  alpha_table4[i] = model$alpha
  sigma_table4[i] = model$sigma2
  shift_values4[[i]]=model$shift.values
  mu_table4[i,]=model$mu
  RE = sqrt_OU_covariance(lizard$tree ,alpha=model$alpha,
                           root.model = "OUfixedRoot",
                           check.order=FALSE, check.ultrametric=FALSE)
  covInverseSqrt <- t(RE$sqrtInvSigma)
  contrast_table4[i,] <- covInverseSqrt%*%(Y_table[i,] - model$mu)
}
save(contrast_table4,file="unknownconfig_contrast_4.1.RData")
save(alpha_table4, file="unknownconfig_alpha_4.1.RData")
save(sigma_table4,file="unknownconfig_sigma2_4.1.RData")
save(mu_table4,file="unknownconfig_mu_4.1.RData")
save(shift_values4,file="unknownconfig_shiftvalues_4.1.RData")
save(vectorOfShift,file="unknownconfig_shifts_4.1.RData")
```

scenario 4.2: unknown shift configuration, unknown alpha and unknown beta

criterion: AICc

```
n_sim=800 # lower because estimation is a lot slower when we have to search for the config
# use first n_sim rows of Ytable only
Y_table=Y_table[1:n_sim,]
contrast_table5=matrix(nrow=n_sim, ncol=n_tips, data=NA)
vectorOfShift2 <- vector(mode = "list", length = n_sim)
alpha_table5 = rep(NA,n_sim)
sigma_table5 = rep(NA,n_sim)
mu_table5 = matrix(nrow=n_sim, ncol=n_tips, data=NA)
shift_values5 = vector(mode = "list", length = n_sim)

for (i in 1:n_sim) {
  model= estimate_shift_configuration(lizard$tree, Y_table[i,],criterion="AICc" )
  vectorOfShift2[[i]] =model$shift.configuration
```

```

alpha_table5[i] = model$alpha
sigma_table5[i] = model$sigma2
shift_values5[[i]]=model$shift.values
mu_table5[i,]=model$mu
RE = sqrt_OU_covariance(lizard$tree ,alpha=model$alpha,
                        root.model = "OUfixedRoot",
                        check.order=FALSE, check.ultrametric=FALSE)

covInverseSqrt <- t(RE$sqrtInvSigma)
contrast_table5[i,] <- covInverseSqrt%*%(Y_table[i,] - model$mu)
}
save(contrast_table5,file="unknownconfig_contrast_4.2.RData")
save(alpha_table5, file="unknownconfig_alpha_4.2.RData")
save(sigma_table5,file="unknownconfig_sigma2_4.2.RData")
save(mu_table5,file="unknownconfig_mu_4.2.RData")
save(shift_values5,file="unknownconfig_shiftvalues_4.2.RData")
save(vectorOfShift2,file="unknownconfig_shifts_4.2.RData")

```

Results: visualizations of contrast distributions

```

#nodes that corresponding to small variance contrasts
intnode=lizard.tree$edge[,1]
all.equal(intnode, rep(199:101, each=2))

```

```
## [1] TRUE
```

```
intnode=unique(intnode) ##remove repetitive numbers
```

The sequence of internal node is from 199:101, which corresponding to contrasts from 1: 99. The 100th contrast is the contrast of the root node. Here is a simple proof: Now to check the mean and variance of the simulated Y values:

```

# Now, we want to know which contrast does node 197 corresponding to. After we add 5 to all traits value
truealpha=eModel$alpha
REf = sqrt_OU_covariance(lizard.tree, alpha=truealpha,
                        root.model = eModel$liou.options$root.model,
                        check.order=F, check.ultrametric=F)

Dtf = t(REf$sqrtInvSigma)
contrast_original=Dtf%*%(eModel$Y - eModel$mu)
eModel$Y[99]=eModel$Y[99]+5
eModel$Y[98]=eModel$Y[98]-5
eModel$Y[97]=eModel$Y[97]-5
eModel$Y[96]=eModel$Y[96]-5
contrast_change=Dtf%*%(eModel$Y - eModel$mu)
options(scipen=999)
cbind(contrast_original,contrast_change)

```

```

##                [,1]                [,2]
##    [1,] -0.10947518720180664 -0.109475187
##    [2,] -0.20391621170386781 -0.203916212
##    [3,] -0.16836339207557977 -12.802737545
##    [4,] -0.17517647154780189 -0.175176472
##    [5,]  0.12732390795686993  0.127323908

```

##	[6,]	0.41444565695355912	0.414445657
##	[7,]	0.25622293765488147	0.256222938
##	[8,]	0.79739857446070006	2.871655463
##	[9,]	-0.10215266190625116	-0.102152662
##	[10,]	-0.07654323764936991	-0.076543238
##	[11,]	0.22266361486011427	0.222663615
##	[12,]	0.29620190282784437	0.296201903
##	[13,]	-0.20014166745501877	0.469654355
##	[14,]	0.24630449148053085	0.246304491
##	[15,]	0.30678236248580520	0.906779820
##	[16,]	-0.05228136002252097	-0.404287138
##	[17,]	-0.01156318144703972	-0.011563181
##	[18,]	-0.10881056113728954	-0.108810561
##	[19,]	-0.82369060763912372	-0.823690608
##	[20,]	0.08883698279294662	0.088836983
##	[21,]	0.24087678680282099	0.240876787
##	[22,]	0.42436466955939178	0.424364670
##	[23,]	0.10123126465911000	0.101231265
##	[24,]	0.13214554643056095	0.132145546
##	[25,]	-0.54857898601102362	-0.548578986
##	[26,]	-0.21719368358609842	-0.217193684
##	[27,]	0.44261460470085257	0.442614605
##	[28,]	0.12302260183133965	0.123022602
##	[29,]	0.12774703852808544	0.127747039
##	[30,]	0.10991519921449291	0.109915199
##	[31,]	0.15269325398011388	0.152693254
##	[32,]	0.01559767597371309	0.015597676
##	[33,]	0.05878371655583753	0.058783717
##	[34,]	0.08219759885015171	0.082197599
##	[35,]	-0.74069790336751351	-0.740697903
##	[36,]	-0.33125218701662390	-0.331252187
##	[37,]	0.06906407801163141	0.069064078
##	[38,]	0.22486210735706463	0.224862107
##	[39,]	0.24455094796942473	0.244550948
##	[40,]	-0.40893176298358963	-0.408931763
##	[41,]	-0.39290292938698346	-0.392902929
##	[42,]	-0.08295851926307238	-0.082958519
##	[43,]	0.06808987686994178	0.068089877
##	[44,]	0.05608603180076910	0.056086032
##	[45,]	-0.14340772870844740	-0.143407729
##	[46,]	-0.79950172014086185	-0.799501720
##	[47,]	-0.12071242724422748	-0.120712427
##	[48,]	-0.00377694651711848	-0.003776947
##	[49,]	0.01982999253016450	0.019829993
##	[50,]	-0.02731047221956251	-0.027310472
##	[51,]	-0.09786195803743299	-0.097861958
##	[52,]	0.11716437857299533	0.117164379
##	[53,]	0.03701845299343412	0.037018453
##	[54,]	0.26251621532315095	0.262516215
##	[55,]	-0.00414884272676316	-0.004148843
##	[56,]	-0.17781139288956221	-0.177811393
##	[57,]	0.07074554554806685	0.070745546
##	[58,]	0.09158298448682092	0.091582984
##	[59,]	-0.00926447502481605	-0.009264475

```
## [60,] 0.32801206751722367 0.328012068
## [61,] -0.07538346472426145 -0.075383465
## [62,] 0.01205077976859600 0.012050780
## [63,] -0.14040997953331524 -0.140409980
## [64,] -0.29119427606946491 -0.291194276
## [65,] 0.0279155668065551 0.027915567
## [66,] 0.21553645531217711 0.215536455
## [67,] -0.19819860043871512 -0.198198600
## [68,] 0.11504496220451804 0.115044962
## [69,] 0.17175157534445609 0.171751575
## [70,] -0.16834246964768601 -0.168342470
## [71,] 0.10221808681558664 0.102218087
## [72,] 0.36538847403961938 0.365388474
## [73,] 0.29408134777332556 0.294081348
## [74,] 0.04586877664551146 0.045868777
## [75,] 0.05392494430993808 0.053924944
## [76,] 0.38314657194311141 0.383146572
## [77,] -0.26727759597524625 -0.267277596
## [78,] 0.23701378566859410 0.237013786
## [79,] -0.20418134660822992 -0.204181347
## [80,] 0.05872596182633311 0.058725962
## [81,] -0.00458795124548232 -0.004587951
## [82,] -0.12782695529349680 -0.127826955
## [83,] -0.37006249194232949 -0.370062492
## [84,] -0.11476953145425853 -0.114769531
## [85,] -0.05706542383790767 -0.057065424
## [86,] -0.01897784543956124 -0.018977845
## [87,] 0.04632514344698074 0.046325143
## [88,] 0.08554927061323926 0.085549271
## [89,] 0.23085318770631477 0.230853188
## [90,] -0.16791845737715250 -0.167918457
## [91,] -0.01151117341378606 -0.011511173
## [92,] 0.07723932701150366 0.077239327
## [93,] 0.09052346301305478 0.090523463
## [94,] -0.02053727613353006 -0.020537276
## [95,] 0.22770115095317778 0.227701151
## [96,] -0.16649238471049044 -0.166492385
## [97,] 0.13075389341821070 0.130753893
## [98,] -0.25361707270714468 -0.253617073
## [99,] 0.14801248520906202 0.148012485
## [100,] -0.0000000003619286 -0.000000000
```

Here, we can see the third contrast changes the most significantly, so the node 197 corresponding to contrast 3.

Results from Scenario 1: all parameters known and set to their true values

```
load("allknown_contrast.RData")
head(colMeans(contrast_table1))
```

```
## [1] 0.0002936259 0.0005437081 -0.0011580854 -0.0014622974 0.0013248416
## [6] -0.0001489877
```



```
max(abs(colMeans(contrast_table1)))# 0.0018224
```

```
## [1] 0.001822459
```

```
round(colMeans(contrast_table1),5)
```

```
## [1] 0.00029 0.00054 -0.00116 -0.00146 0.00132 -0.00015 -0.00011
## [8] 0.00032 0.00109 -0.00039 -0.00069 -0.00036 -0.00044 0.00150
## [15] 0.00060 0.00015 -0.00048 0.00109 -0.00112 0.00032 -0.00021
## [22] 0.00124 0.00055 0.00019 -0.00014 0.00166 -0.00067 -0.00171
## [29] -0.00113 0.00035 0.00039 -0.00005 0.00079 -0.00073 -0.00058
## [36] 0.00024 0.00016 0.00078 0.00136 -0.00034 -0.00106 -0.00081
## [43] 0.00090 0.00036 -0.00182 -0.00073 -0.00113 0.00001 0.00079
## [50] -0.00036 0.00092 -0.00088 -0.00041 -0.00033 0.00038 -0.00038
## [57] -0.00080 -0.00008 0.00013 -0.00045 0.00070 -0.00048 0.00063
## [64] -0.00121 -0.00094 0.00022 -0.00029 -0.00129 -0.00058 0.00018
## [71] 0.00028 -0.00132 -0.00082 0.00006 -0.00080 -0.00120 0.00040
## [78] 0.00006 0.00045 0.00045 -0.00039 0.00005 0.00058 0.00101
## [85] 0.00033 0.00005 0.00086 0.00040 -0.00028 0.00032 -0.00049
## [92] 0.00142 -0.00022 -0.00119 0.00075 0.00029 -0.00035 -0.00123
## [99] -0.00040 -0.00026
```

```
convar=apply(contrast_table1,2,ss)
```

```
round(convar,3)
```

```
## [1] 0.062 0.063 0.062 0.062 0.062 0.062 0.062 0.063 0.062 0.062 0.062 0.062
## [12] 0.062 0.063 0.062 0.063 0.063 0.063 0.063 0.062 0.062 0.063 0.062 0.063
## [23] 0.063 0.062 0.062 0.063 0.062 0.062 0.062 0.063 0.063 0.062 0.062 0.063
## [34] 0.063 0.063 0.063 0.063 0.063 0.063 0.063 0.063 0.063 0.062 0.062 0.062
## [45] 0.062 0.062 0.062 0.062 0.063 0.062 0.063 0.063 0.062 0.062 0.062 0.063
## [56] 0.062 0.062 0.062 0.062 0.063 0.063 0.063 0.062 0.063 0.063 0.063 0.063
## [67] 0.062 0.063 0.062 0.063 0.063 0.063 0.063 0.062 0.062 0.062 0.062 0.063
## [78] 0.062 0.063 0.063 0.062 0.063 0.063 0.063 0.063 0.063 0.063 0.062 0.062
## [89] 0.063 0.062 0.062 0.063 0.063 0.062 0.062 0.063 0.062 0.063 0.063 0.062
## [100] 0.062
```

```
mean(convar)# 0.06249321
```

```
## [1] 0.06249321
```

```
sigma2# 0.06251866
```

```
## [1] 0.06251866
```

```
0.1089751/0.06251866 # 1.743081. liou v1.29 without normalize.tree.height: mean(convar) was 0.1089751
```

```
## [1] 1.743081
```

Mean of contrasts are close to zero and the variance of contrasts is close to `sigma2`.

Results from Scenario 2: ## scenario 2: beta unknown

but known alpha and known shift configuration

```
load("knownalpha_contrast.RData")
load("knownalpha_sigma2.RData")
load("knownalpha_shiftvalues.RData")
load("knownalpha_mu.RData")
```

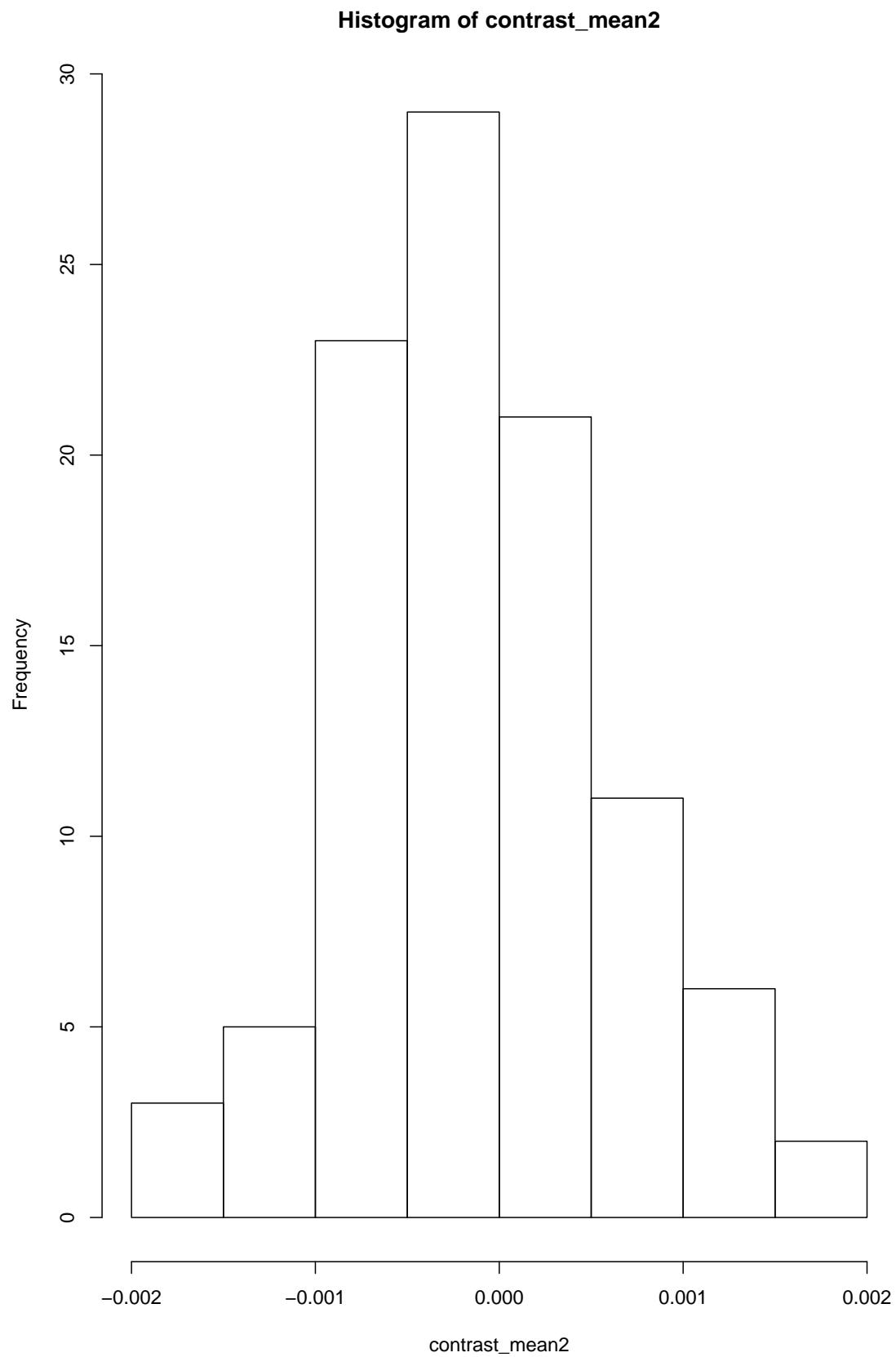
```
# contrasts mean
contrast_mean2=colMeans(contrast_table2)
head(contrast_mean2)

## [1] -0.0008017153 -0.0010609915 -0.0008243967 -0.0009007101 -0.0001081647
## [6]  0.0006750217

max(abs(contrast_mean2))

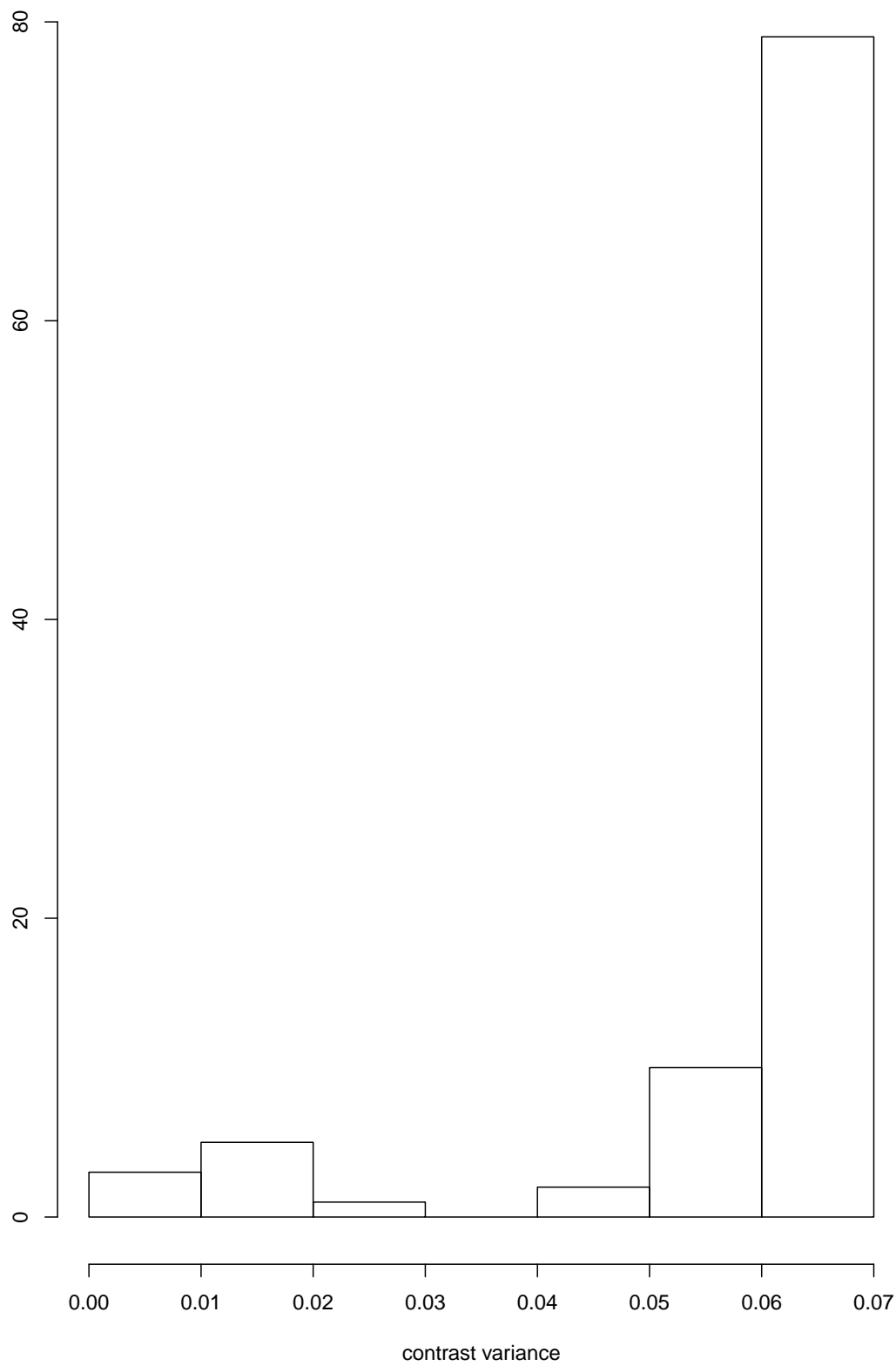
## [1] 0.001775369

hist(contrast_mean2)
```



```
# contrasts variance
convar2=apply(contrast_table2,2,ss)
contrast2=colMeans(contrast_table2)
hist(convar2,main="variance of contrasts in Scenario 2",xlab="contrast variance",ylab="")
```

variance of contrasts in Scenario 2

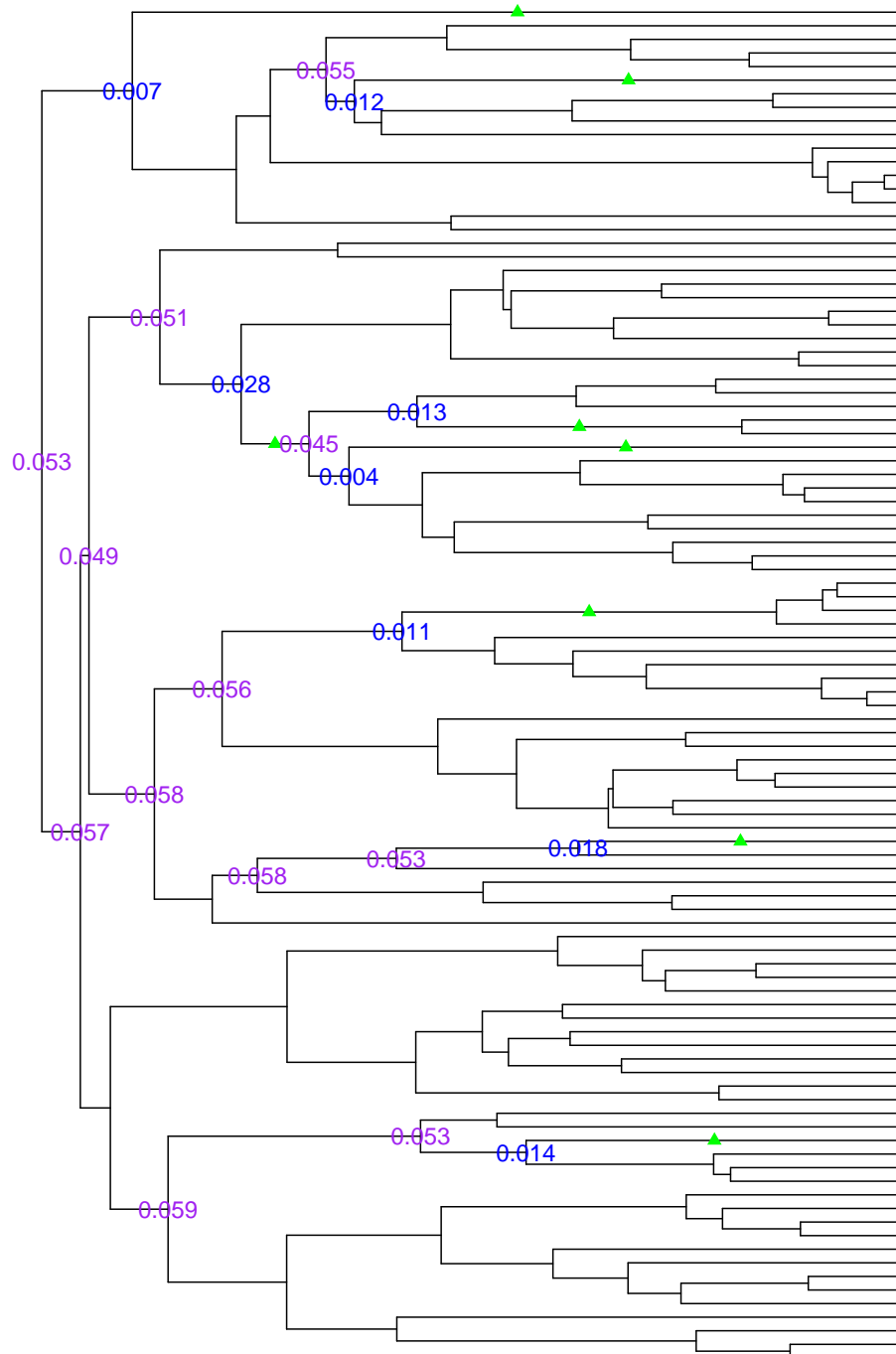


```

#Try to find the contrast with variance relatively small, median and large.
ind_small2=which(convar2<0.04)
ind_median2=which(convar2<0.06&convar2>=0.04)
median_node2=intnode[ind_median2]
small_node2=intnode[ind_small2]
plot(eModel$tree,show.tip.label = F,main="",font=8)
nodelabels(round(convar2[ind_small2],3),small_node2,col="blue",frame = "none")
edgelabels(edge=shift_config,col="green",frame = "none",pch=17)
nodelabels(round(convar2[ind_median2],3),median_node2,col="purple",frame = "none")
mtext("blue: contrast variance < 0.04, purple: contrast variance > 0.04 and < 0.06,
      green dots: shifts",
      line=1,side=3,las=0)

```

blue: contrast variance < 0.04, purple: contrast variance > 0.04 and < 0.06,
green dots: shifts



```
round(convar2,3)
```

```
## [1] 0.062 0.062 0.062 0.063 0.063 0.063 0.012 0.055 0.063 0.063 0.062
## [12] 0.063 0.061 0.063 0.061 0.007 0.063 0.062 0.062 0.062 0.062 0.063
## [23] 0.062 0.062 0.063 0.063 0.062 0.013 0.063 0.063 0.063 0.063 0.062
## [34] 0.063 0.062 0.063 0.004 0.045 0.028 0.051 0.063 0.062 0.063 0.063
## [45] 0.062 0.063 0.063 0.062 0.011 0.062 0.062 0.062 0.062 0.062 0.062
## [56] 0.063 0.063 0.056 0.018 0.053 0.063 0.063 0.058 0.061 0.058 0.049
## [67] 0.063 0.062 0.063 0.063 0.063 0.062 0.062 0.062 0.063 0.063 0.063
## [78] 0.063 0.063 0.063 0.063 0.014 0.053 0.063 0.063 0.062 0.063 0.063
## [89] 0.063 0.063 0.062 0.063 0.063 0.063 0.062 0.059 0.062 0.057 0.053
## [100] 0.000
```

```
mean(convar2)
```

```
## [1] 0.0569135
```

```
mean(convar2[-ind_small2])
```

```
## [1] 0.06136439
```

```
sigma2
```

```
## [1] 0.06251866
```

Some contrasts with extremely small variance draw our attention. When we make a plot, we can see those contrasts with small variances appear where the shift exists. Those shifts make the variances of contrasts underestimated. And the plot below can clearly show as nodes become further away from shifts, their variances of contrast increase. The nodes closer to the root have higher variance of contrasts. And it is clear that the last contrast has variance equal to 0 since we have a fixed root.

```
###making plots for contrasts
```

```
dist2shift=rep(Inf,199)
```

```
dist2shift[shiftnode]=0
```

```
shiftparent=shiftnode
```

```
for(k in 1:10){
```

```
  old=c()
```

```
  for(node in shiftparent){
```

```
    ind=which(node==eModel$tree$edge[,2])
```

```
    old=c(old,ind)
```

```
  }
```

```
  shiftparent=eModel$tree$edge[old,1]
```

```
  for(node in shiftparent){
```

```
    if(dist2shift[node]>k) dist2shift[node]=k
```

```
  }
```

```
}
```

```
dist2shift=dist2shift[-c(1:99)]
```

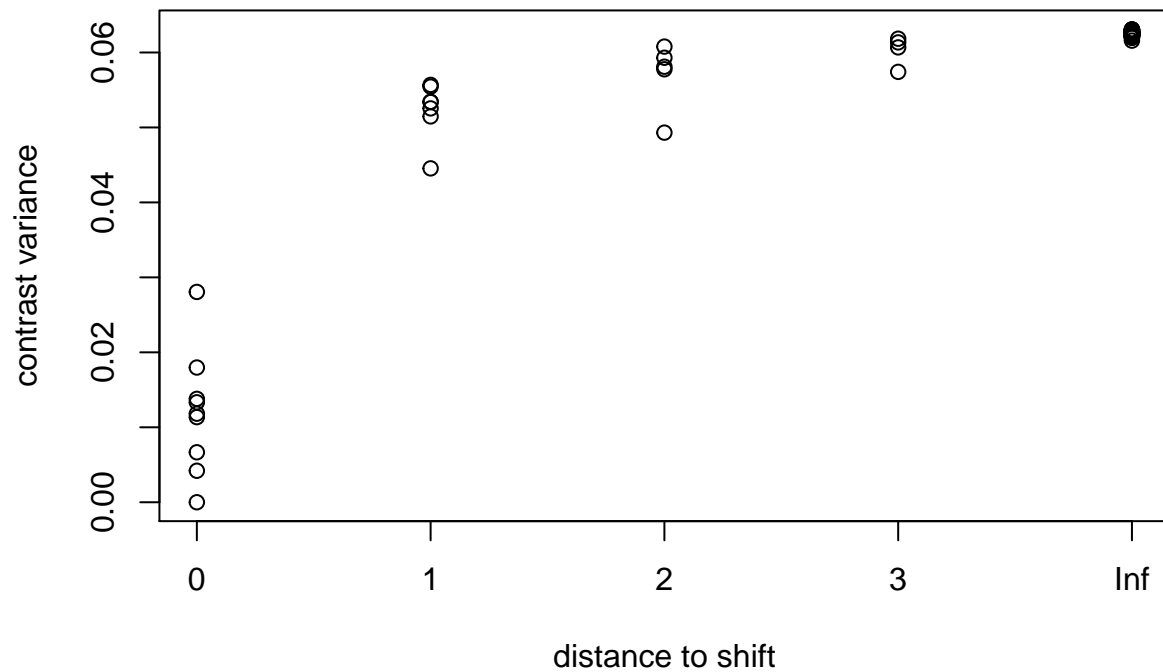
```
dist2shift=rev(dist2shift)
```

```
dist2shift[100]=0 ##The last contrast is the root edge
```

```
dist2shift[dist2shift==Inf] = 4
```

```
plot(convar2~dist2shift, xaxt="n",xlab="distance to shift", ylab="contrast variance")
```

```
axis(1, at=0:4, labels=as.character(c(0:3,Inf)))
```

Results from Scenario 3: alpha and beta unknown

but known shift configuration

```
load("unknownalpha_contrast.RData")
load("unknownalpha_sigma2.RData")
load("unknownalpha_shiftvalues.RData")
load("unknownalpha_mu.RData")
load("unknownalpha_alpha.RData")
# calculate contrasts mean
contrast_mean3=colMeans(contrast_table3)
head(contrast_mean3)
```

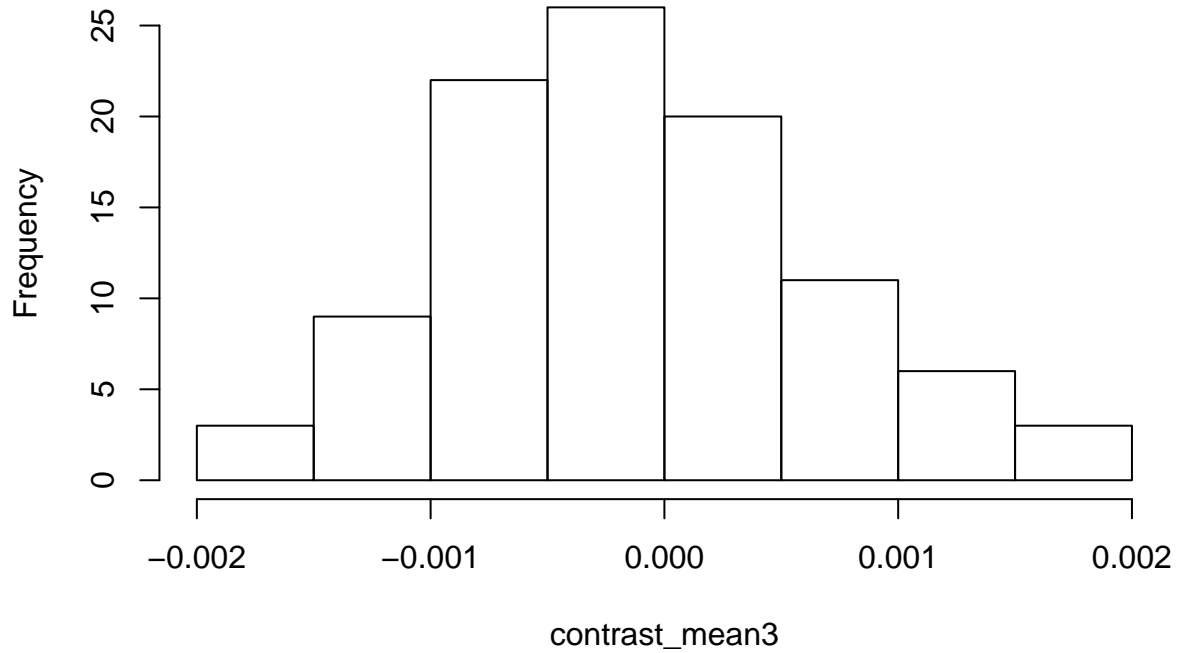
```
## [1] -0.0008208711 -0.0010534489 -0.0008314135 -0.0009439863 -0.0001392928
## [6]  0.0007259835
```

```
max(abs(contrast_mean3))
```

```
## [1] 0.001924415
```

```
hist(contrast_mean3)
```

Histogram of contrast_mean3



```
# Calculate contrasts variance
```

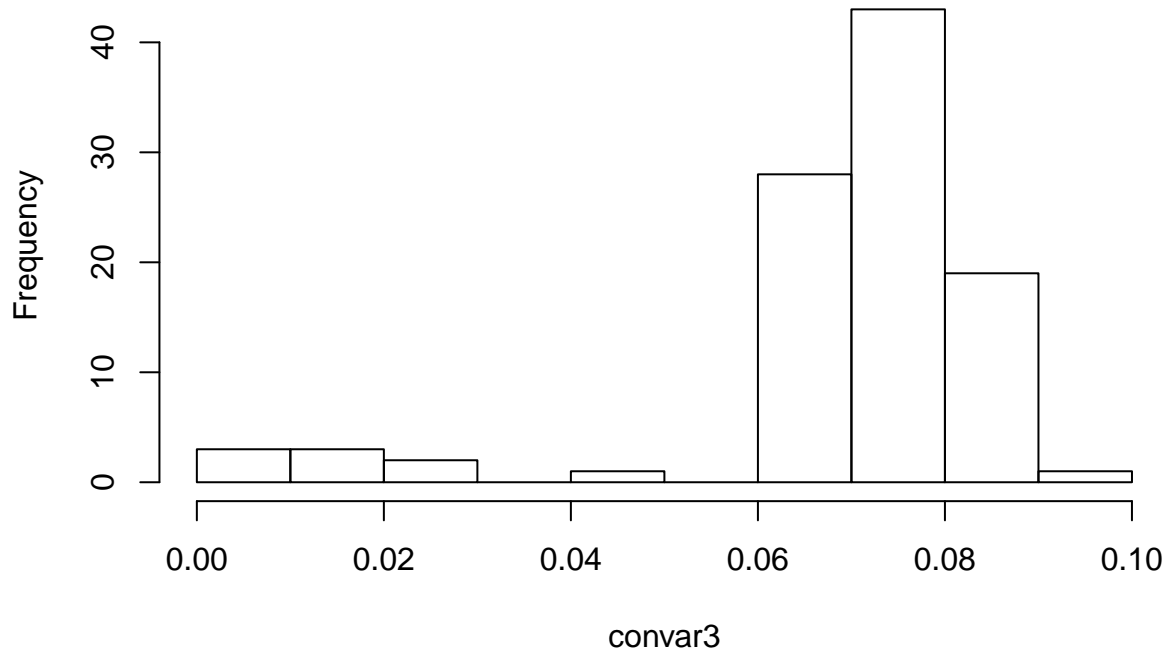
```
convar3=apply(contrast_table3,2,ss)
```

```
round(convar3,3)
```

```
## [1] 0.069 0.074 0.080 0.068 0.076 0.081 0.017 0.076 0.064 0.065 0.066
## [12] 0.067 0.083 0.078 0.084 0.009 0.081 0.071 0.065 0.074 0.079 0.077
## [23] 0.067 0.081 0.070 0.076 0.069 0.020 0.067 0.068 0.076 0.073 0.068
## [34] 0.072 0.082 0.084 0.005 0.062 0.045 0.074 0.066 0.066 0.069 0.064
## [45] 0.066 0.073 0.076 0.078 0.019 0.071 0.067 0.069 0.071 0.075 0.074
## [56] 0.080 0.080 0.080 0.027 0.067 0.072 0.079 0.078 0.082 0.083 0.068
## [67] 0.069 0.072 0.073 0.076 0.075 0.074 0.072 0.078 0.080 0.070 0.083
## [78] 0.089 0.077 0.070 0.070 0.020 0.070 0.067 0.070 0.071 0.067 0.072
## [89] 0.074 0.077 0.083 0.067 0.071 0.081 0.087 0.085 0.092 0.084 0.080
## [100] 0.000
```

```
hist(convar3)
```

Histogram of convar3



```
###Exactly the same as last one
intnode[convar3<0.06]
```

```
## [1] 193 184 172 163 161 151 141 118 NA
```

```
mean(convar3)
```

```
## [1] 0.06909039
```

```
sigma2
```

```
## [1] 0.06251866
```

```
#Try to find the contrast with variance relatively small, median and large.
```

```
ind_small3=which(convar3<0.04)
```

```
ind_median3=which(convar3<0.06&convar3>=0.04)
```

```
ind_large3=which(convar3>=0.08)
```

```
median_node3=intnode[ind_median3]
```

```
small_node3=intnode[ind_small3]
```

```
large_node3=intnode[ind_large3]
```

```
large_node3
```

```
## [1] 194 187 185 183 176 165 164 144 143 136 135 125 123 122 109 106 105
```

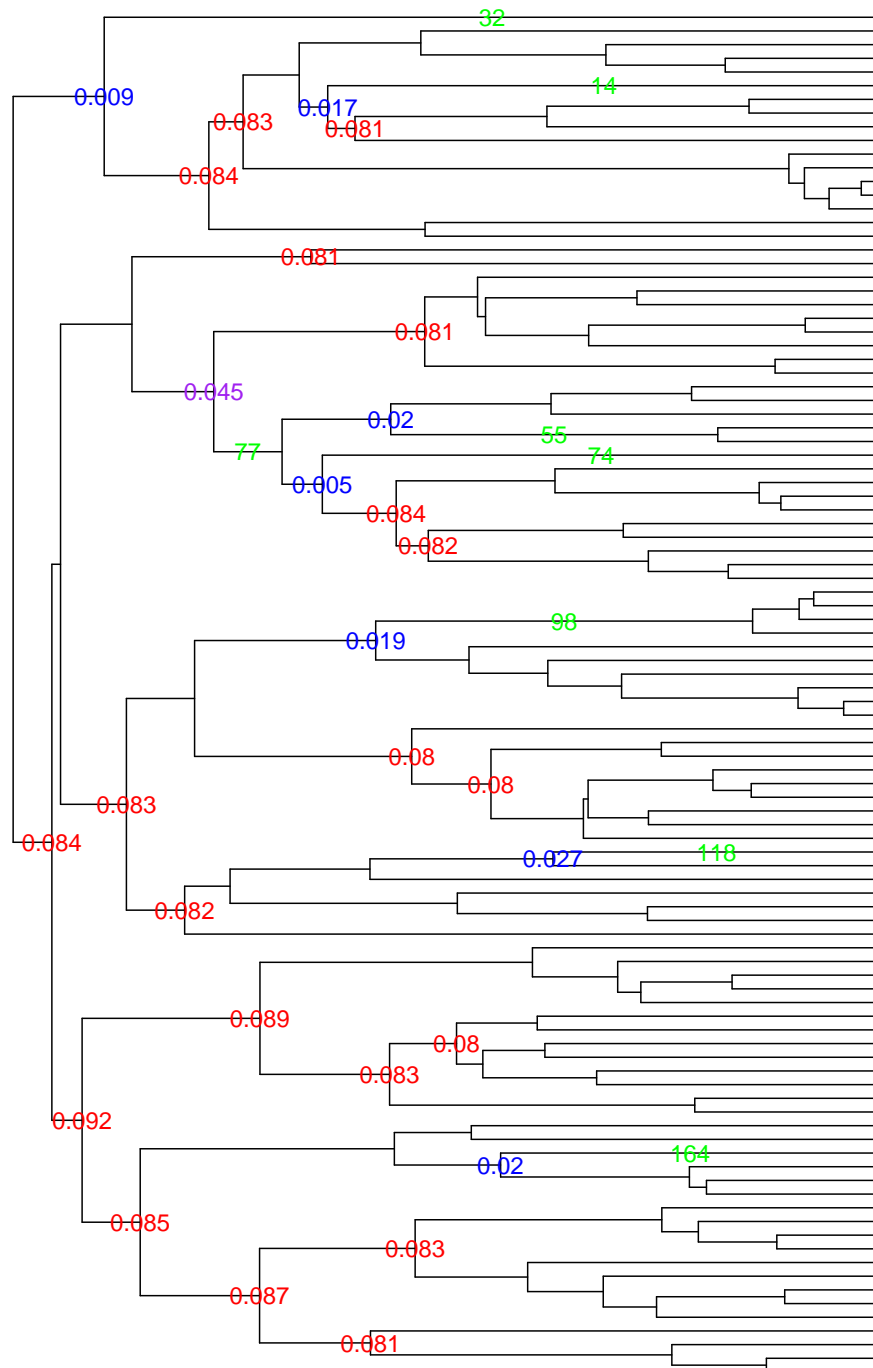
```
## [18] 104 103 102
```

```
mean(convar3[-c(ind_small3,ind_large3)])
```

```
## [1] 0.07117836
```

```
plot(eModel$tree,show.tip.label = F,main="small,median,large contrast nodes",font=8)
nodelabels(round(convar3[ind_small3],3),small_node3,col="blue",frame = "none")
edgelabels(shift_config,shift_config,col="green",frame = "none")
nodelabels(round(convar3[ind_median3],3),median_node3,col="purple",frame = "none")
nodelabels(round(convar3[ind_large3],3),large_node3,col="red",frame = "none")
```

small,median,large contrast nodes



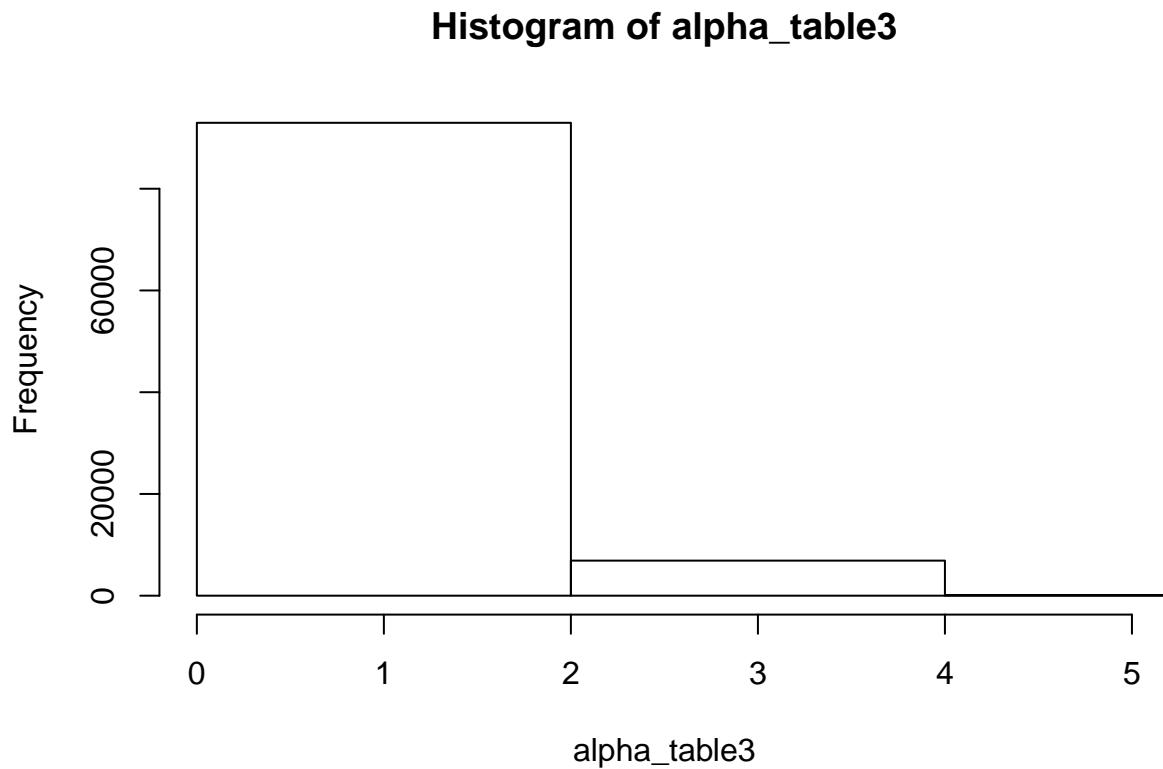
Since al-

pha is unknown, the variance of contrasts being estimated would be different with varied alpha values. So, I split contrast tables into two different sets of contrasts based on either alpha has been underestimated or overestimated.

```
#explore alpha  
n_sim=100000  
mean(alpha_table3)
```

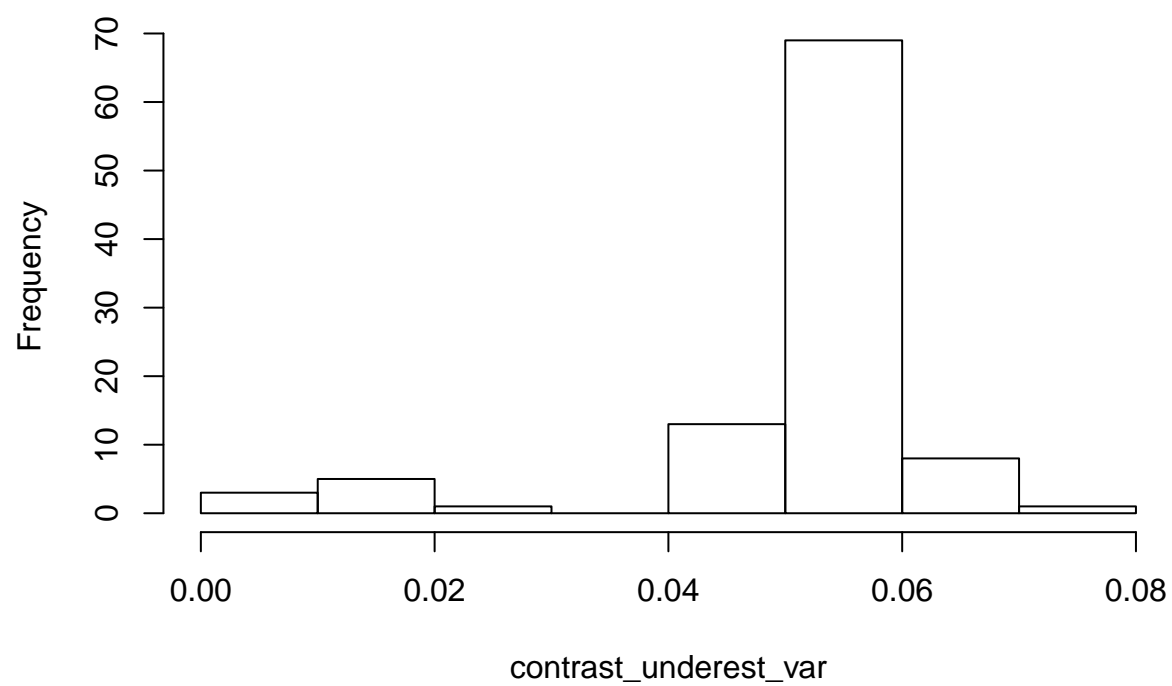
```
## [1] 1.065242
```

```
hist(alpha_table3,xlim=c(0,5))
```



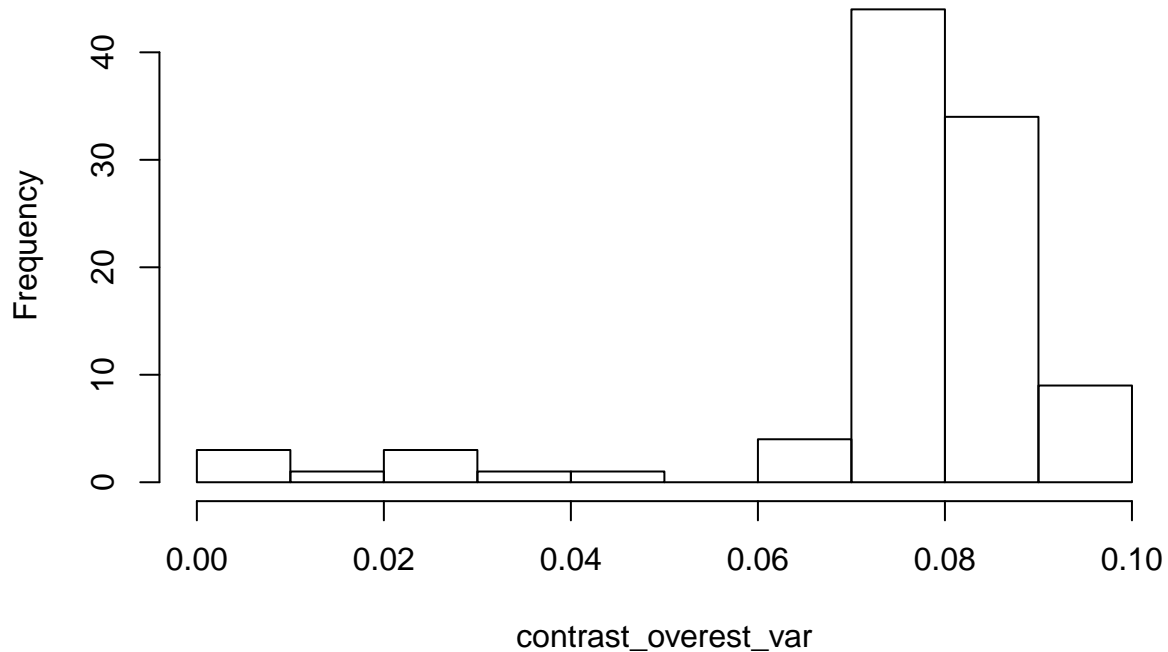
```
alpha_underest=rep(2,n_sim)  
alpha_underest[alpha_table3<eModel$alpha]=1  
contrast_underest=subset(contrast_table3,alpha_underest==1)  
contrast_overest=subset(contrast_table3,alpha_underest==2)  
contrast_underest_var=apply(contrast_underest,2,var)  
contrast_overest_var=apply(contrast_overest,2,var)  
hist(contrast_underest_var)
```

Histogram of contrast_underest_var



```
hist(contrast_overest_var)
```

Histogram of contrast_overest_var



#Try to find the contrast with variance relatively small and large with underestimated alpha

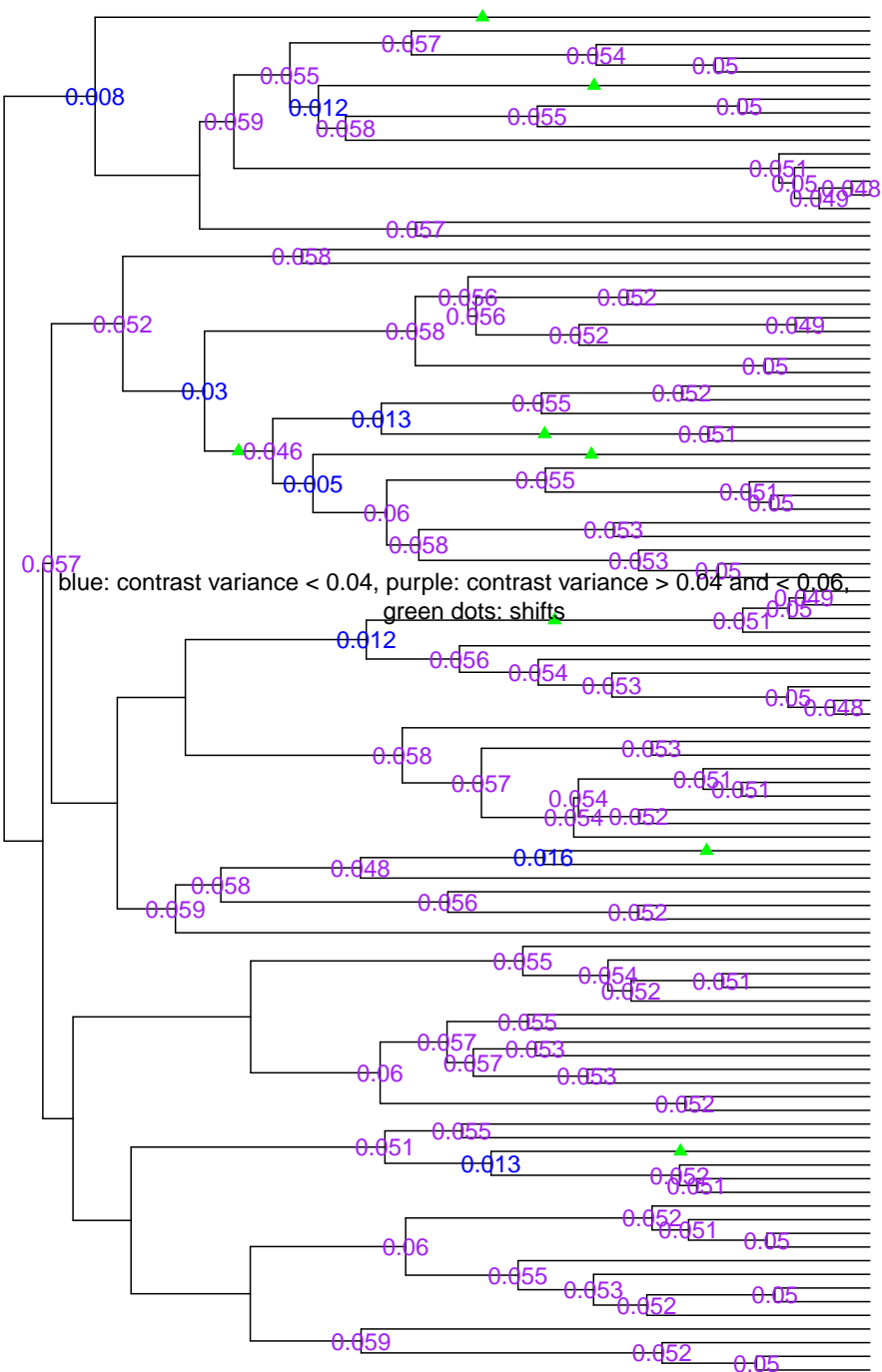
```
ind_small_under=which(contrast_underest_var<0.04)
ind_median_under=which(contrast_underest_var<0.06&contrast_underest_var>=0.04)
ind_large_under=which(contrast_underest_var>=0.08)
median_node3_under=intnode[ind_median_under]
small_node3_under=intnode[ind_small_under]
large_node3_under=intnode[ind_large_under]
large_node3_under
```

```
## integer(0)
```

Try to find the contrast's variance with underestimated alpha

```
plot(eModel$tree,show.tip.label = F,main="estimated alpha < true alpha",font=8)
nodelabels(round(contrast_underest_var[ind_small_under],3),small_node3_under,col="blue",frame = "none")
edgelabels(edge=shift_config,col="green",frame = "none",pch=17)
nodelabels(round(contrast_underest_var[ind_median_under],3),median_node3_under,col="purple",frame = "none")
##nodelabels(round(contrast_underest_var[ind_large_under],3),large_node3_under,col="red",frame = "none")
mtext("blue: contrast variance < 0.04, purple: contrast variance > 0.04 and < 0.06,
      green dots: shifts",
      line=-23,side=3,las=0)
```


estimated alpha < true alpha



Many

contrasts with at least one child node as tip of the tree are underestimated. As mentioned above, contrasts of nodes with a shift edge are underestimated. One of the node labled in red has relatively large variance of contrast. This situation cannot be fully explained by its location closer to the root. (need some investigations)

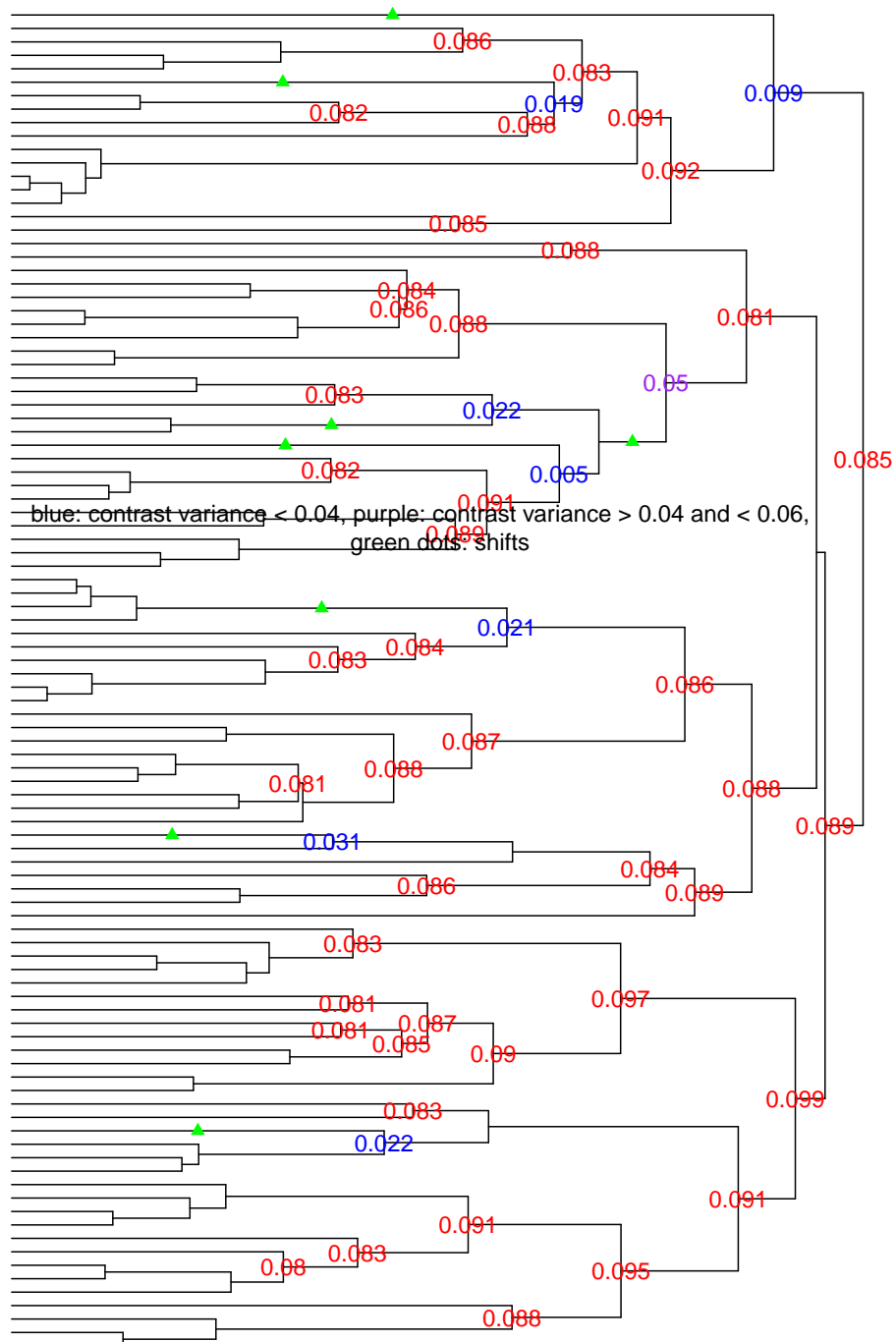
Try to find the contrast's variance with overestimated alpha

```
ind_small_over=which(contrast_overest_var<0.04)
ind_median_over=which(contrast_overest_var<0.06&contrast_overest_var>=0.04)
ind_large_over=which(contrast_overest_var>=0.08)
median_node3_over=intnode[ind_median_over]
small_node3_over=intnode[ind_small_over]
large_node3_over=intnode[ind_large_over]
large_node3_over
```

```
## [1] 197 195 194 192 187 186 185 183 179 178 176 174 169 165 164 160 153
## [18] 152 146 144 143 142 138 137 136 135 130 129 128 126 125 123 122 121
## [35] 111 110 109 106 105 104 103 102 101
```

```
plot(eModel$tree,show.tip.label = F,main="estimated alpha > true alpha",font=8,direction = "leftwards")
nodelabels(round(contrast_overest_var[ind_small_over],3),small_node3_over,col="blue",frame = "none")
edgelabels(edge=shift_config,col="green",frame = "none",pch=17)
nodelabels(round(contrast_overest_var[ind_median_over],3),median_node3_over,col="purple",frame = "none")
nodelabels(round(contrast_overest_var[ind_large_over],3),large_node3_over,col="red",frame = "none")
mtext("blue: contrast variance < 0.04, purple: contrast variance > 0.04 and < 0.06,
      green dots: shifts",
      line=-21,side=3,las=0)
```

estimated alpha > true alpha



When

alpha has been overestimated, there are much more nodes with large variance of contrasts. And most of those nodes are closer to root.

Results from unknown shift configuration

and also unknown alpha and unknown beta. Spent 24hrs, generated 1262 eModels

```
load("unknownconfig_alpha_4.1.RData")
load("unknownconfig_shifts_4.1.RData")
load("unknownconfig_sigma2_4.1.RData")
load("unknownconfig_mu_4.1.RData")
load("unknownconfig_shiftvalues_4.1.RData")
load("unknownconfig_contrast_4.1.RData")
#Calculate mean of contrasts
contrast_mean4=colMeans(contrast_table4)
head(contrast_mean4)

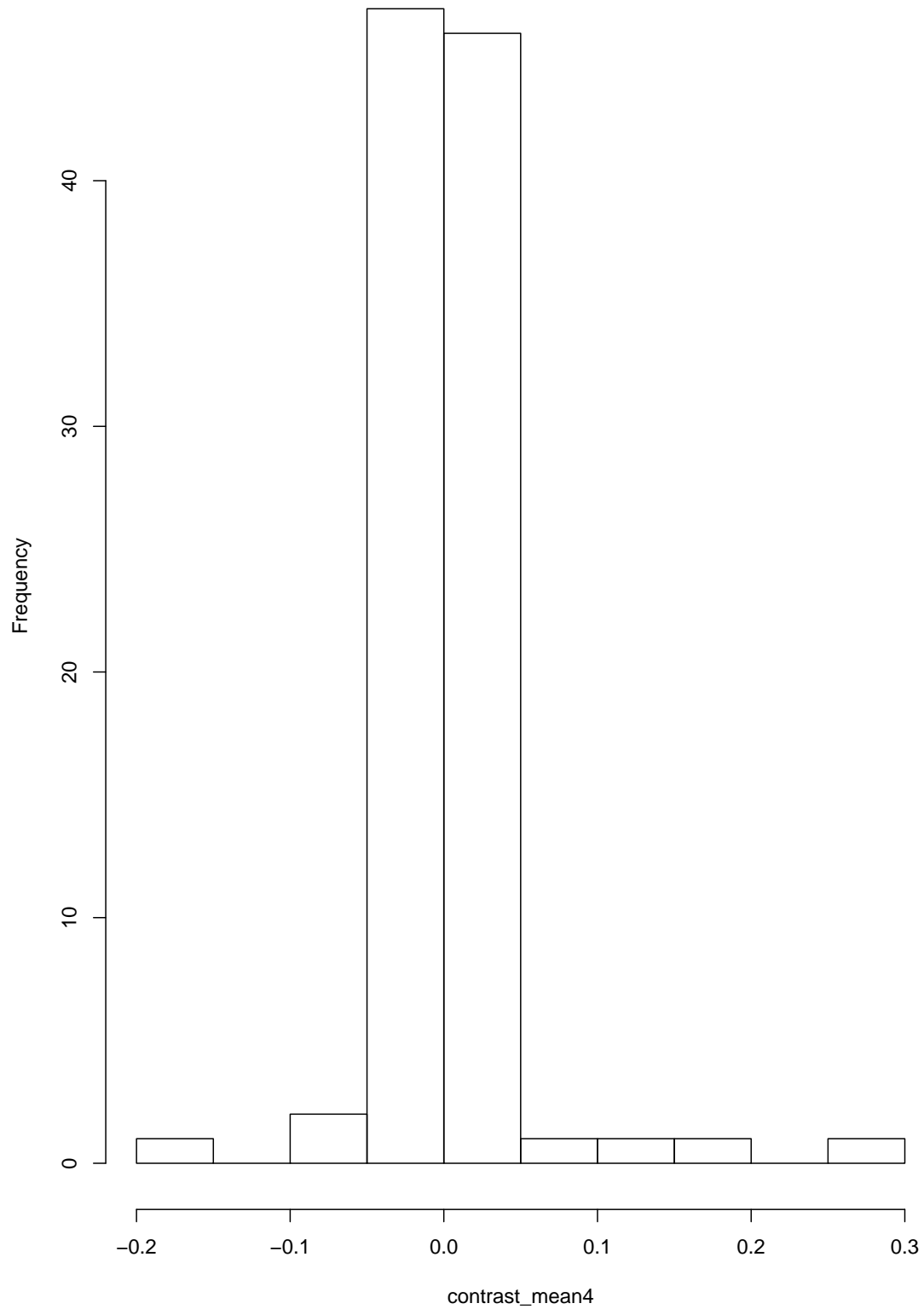
## [1] -0.0107097344 -0.0004473309 -0.0027052034 -0.0142488107 0.0026654166
## [6] -0.0041520054

max(abs(contrast_mean4))

## [1] 0.2764788

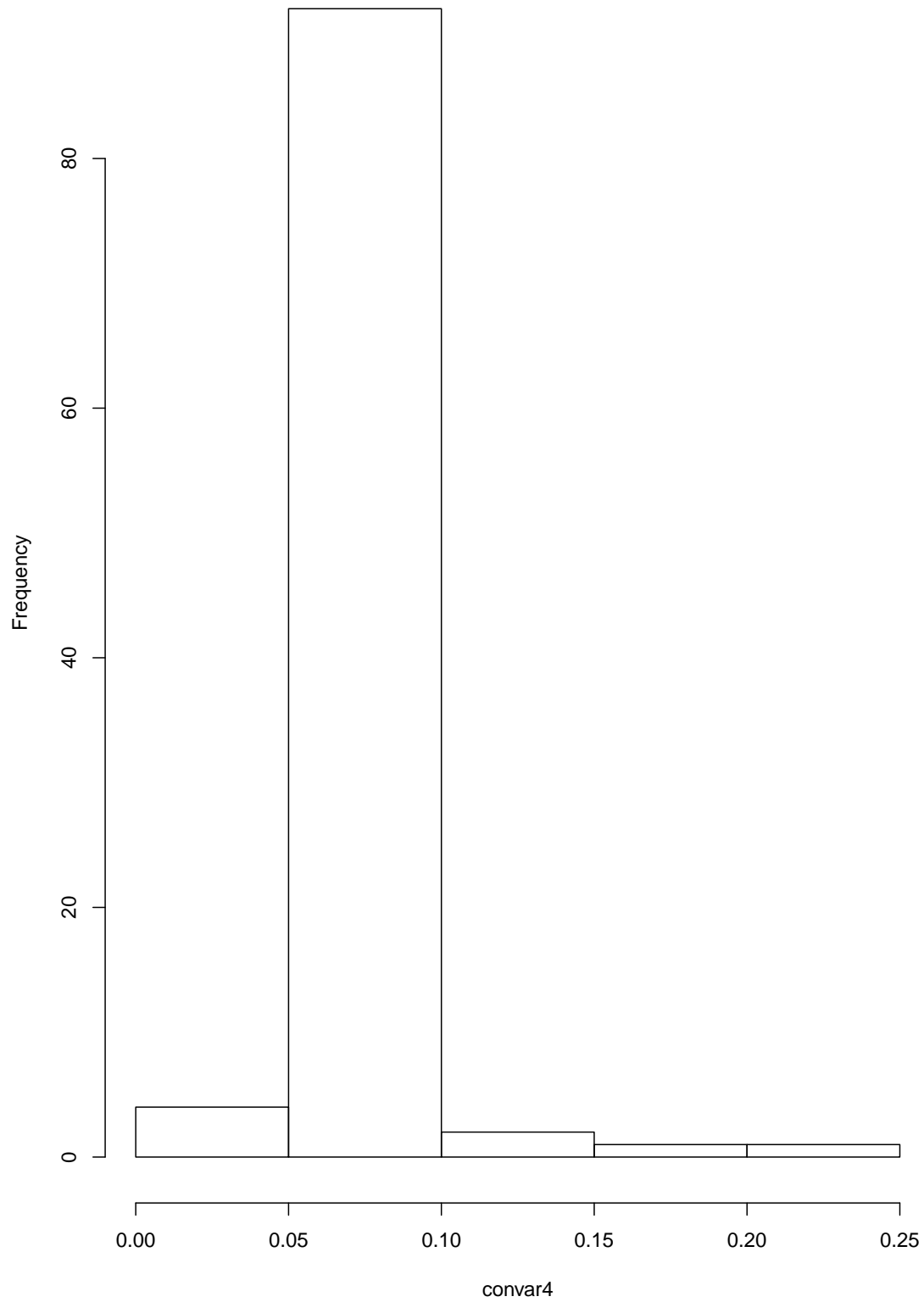
hist(contrast_mean4)
```

Histogram of contrast_mean4



```
#Calculate variance of contrasts  
convar4=apply(contrast_table4,2,ss)  
hist(convar4)
```

Histogram of convar4



```
#Shifts that occur the most often
vectorOfShift_1=unlist(vectorOfShift, recursive = TRUE, use.names = F)
freq_table=as.data.frame(table(vectorOfShift_1))
freq_table
```

```
##      vectorOfShift_1 Freq
## 1              1      1
## 2             14    625
## 3             15      2
## 4             26      1
## 5             27      1
## 6             32    799
## 7             43      1
## 8             52      1
## 9             53      1
## 10            54      1
## 11            55    624
## 12            56     15
## 13            70      2
## 14            71      5
## 15            72      2
## 16            73     21
## 17            74    495
## 18            77    770
## 19            79      3
## 20            83      1
## 21            89      1
## 22            95      2
## 23            98    730
## 24           107      1
## 25           112      1
## 26           118    793
## 27           120      2
## 28           122      1
## 29           124      2
## 30           132      1
## 31           135      1
## 32           137      1
## 33           138      1
## 34           141      1
## 35           163      1
## 36           164    667
## 37           165      2
## 38           173      1
## 39           179      1
## 40           191      1
## 41           192      1
```

Try to find the contrast with variance relatively small, median and large.

```
ind_small4=which(convar4<0.04)
ind_median4=which(convar4<0.06&convar4>=0.04)
ind_large4=which(convar4>=0.085)
median_node4=intnode[ind_median4]
small_node4=intnode[ind_small4]
```



```
large_node4=intnode[ind_large4]
large_node4
```

```
## [1] 193 192 172 163 118
```

```
mean(convar4[-as.numeric(large_node4)])
```

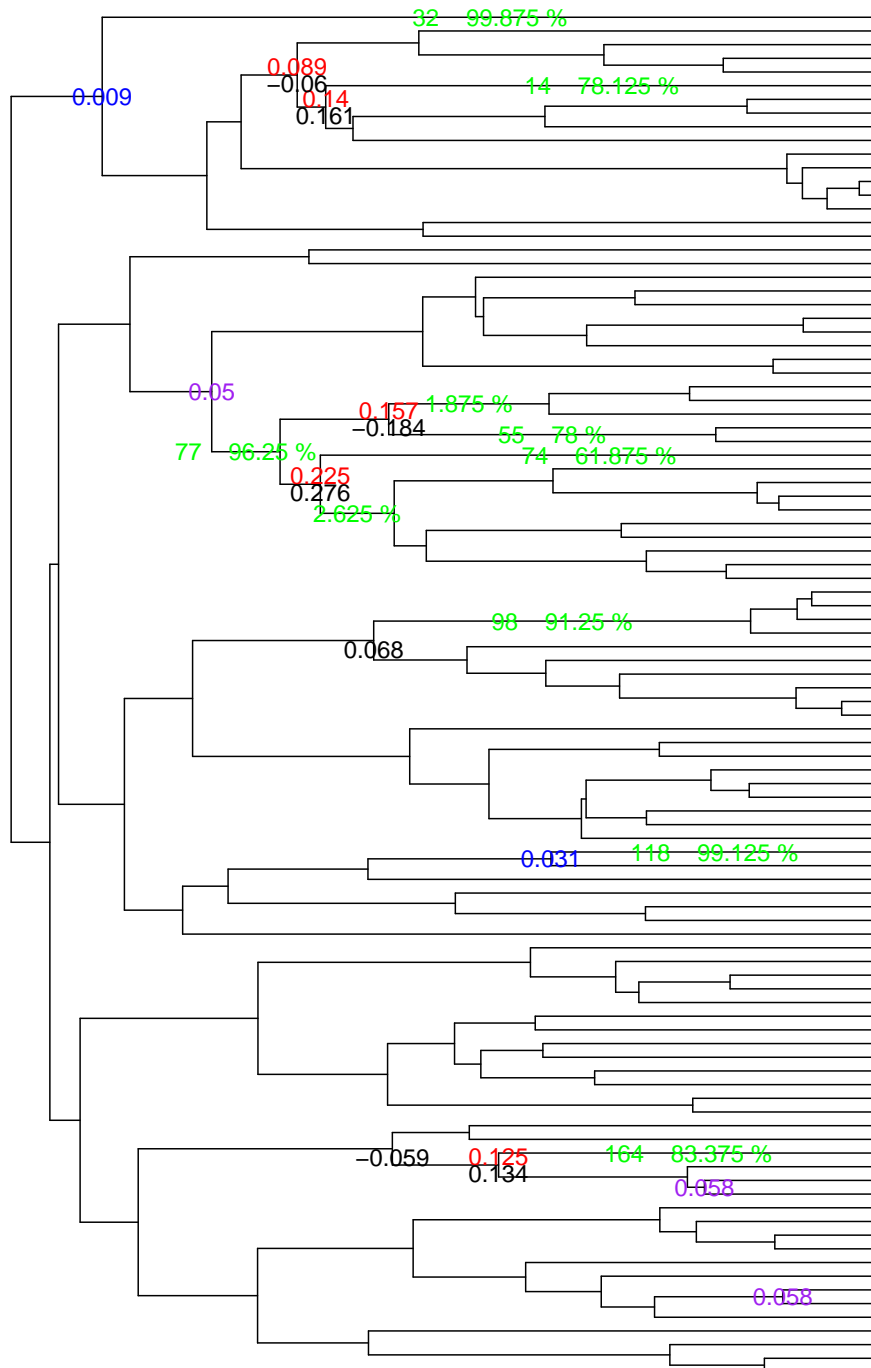
```
## [1] 0.07022559
```

Even though we ignore those nodes with large variance of contrasts, the variances of contrasts on average are still overestimated.

```
mean_large4=intnode[which(abs(contrast_mean4)>0.05)]
median_node4=intnode[ind_median4]
small_node4=intnode[ind_small4]
large_node4=intnode[ind_large4]
plot(eModel$tree,show.tip.label = F,main="small,median,large contrast nodes",font=8)
nodelabels(round(convar4[ind_small4],3),small_node4,col="blue",frame = "none")
for(i in 1:length(shift_config)){
  edgelabels(paste(shift_config[i],"  ",round(freq_table$Freq[which(freq_table$vectorOfShift_1==shift_con
}
#56,73

edgelabels(paste(round(freq_table$Freq[which(freq_table$vectorOfShift_1==56)]/8,3),"%"),56,col="green",
edgelabels(paste(round(freq_table$Freq[which(freq_table$vectorOfShift_1==73)]/8,3),"%"),73,col="green",
nodelabels(round(convar4[ind_median4],3),median_node4,col="purple",frame = "none")
nodelabels(round(convar4[ind_large4],3),large_node4,col="red",frame = "none",adj=c(0.5,0))
nodelabels(round(contrast_mean4[which(abs(contrast_mean4)>0.05)],3),mean_large4,col="black",frame = "none")
```

small,median,large contrast nodes



The

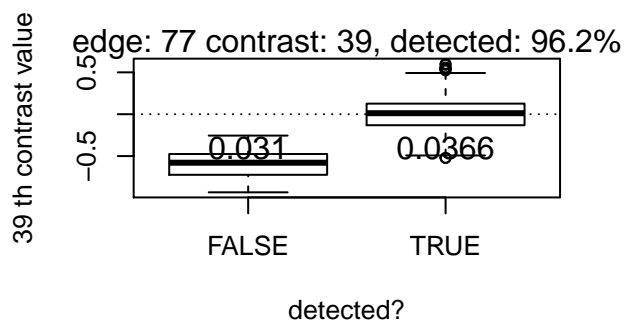
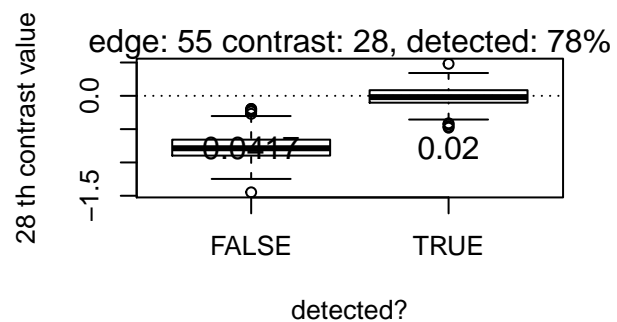
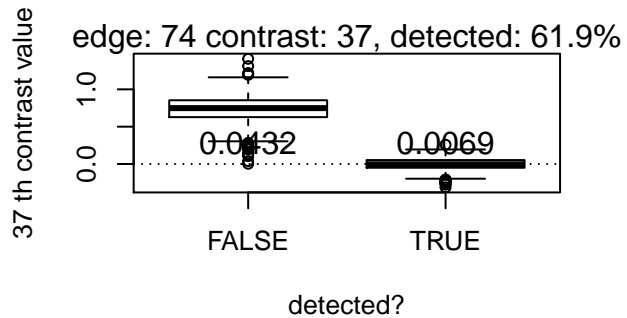
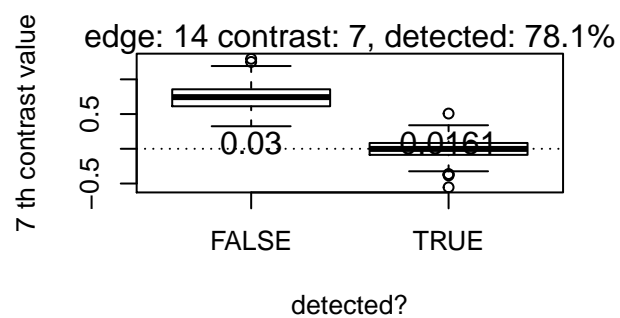
shifts being detected most often are: 32,77,118,164,98,14,55,74 which are exactly corresponding to the shift configuration 55, 98, 118, 74, 14, 77, 32, 164 of the true model. A few nodes with shift edges have larger than average mean of contrasts. So, their variances are overestimated accordingly.

Contrasts of node with shift edges being detected or not

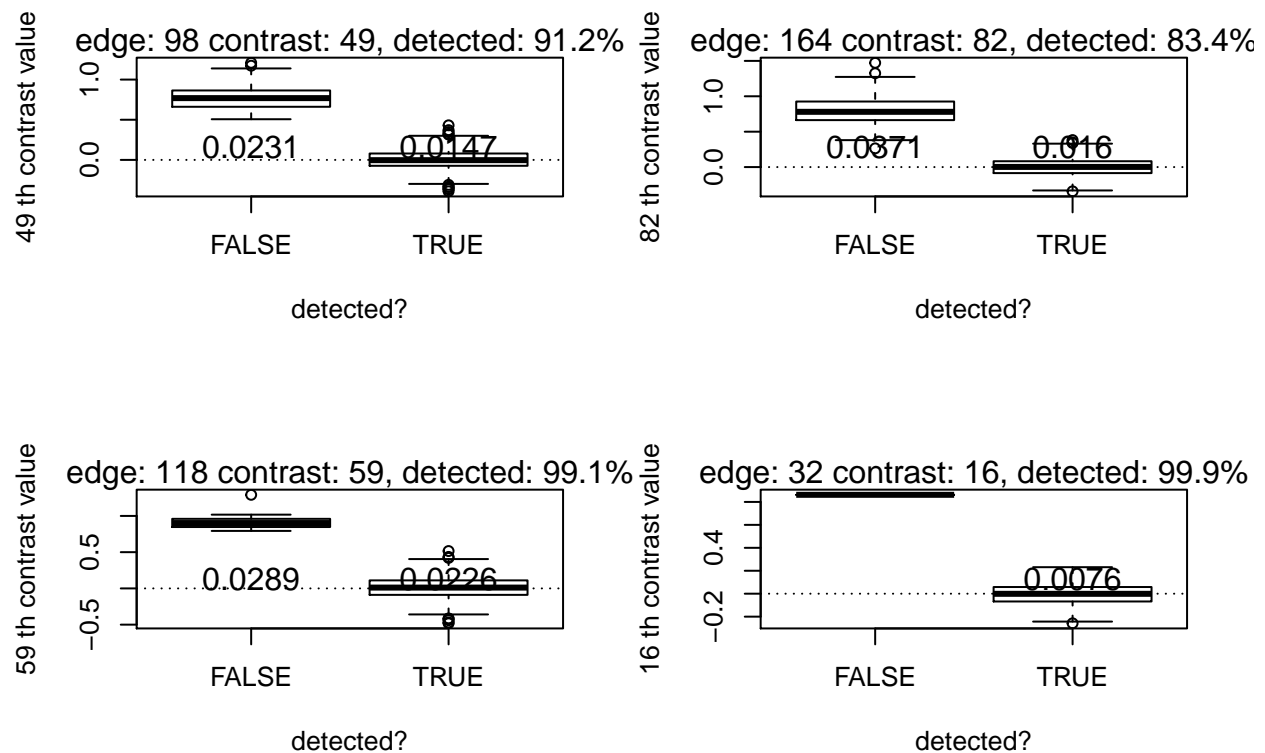
```
contrast_plot = function(edge, contrast){
  contrast_detect=rep(NA, 600)
  for(i in 1:length(vectorOfShift)){
    if(edge%in%vectorOfShift[[i]]){
      contrast_detect[i]=TRUE
    }
    else {
      contrast_detect[i]=FALSE
    }
  }
  plot(contrast_table4[,contrast] ~ factor(contrast_detect), xlab="detected?", ylab=paste(contrast, "th c
  mtext(paste("edge: ", edge, " contrast: ", contrast,
    ", detected: ", round(100*sum(contrast_detect)/800, 1), "%", sep=""))
  abline(h=0, lty=3)
  convar_bygroup = tapply(contrast_table4[,contrast], contrast_detect, var)
  mtext(side=1, at=1:2, text=round(convar_bygroup,4), line=-2)
}
```

Those figures show below represent contrasts of different nodes with group1(shift detected) and group2(shift not detected).

```
layout(matrix(1:4,2,2))
contrast_plot(14,7) # edge 14, node 193, contrast 7
contrast_plot(55,28) # edge 55, node 172, contrast 28
contrast_plot(74,37) # edge 74, node 163, contrast 37
contrast_plot(77,39) # edge 77, node 161, contrast 39
```



```
contrast_plot(98,49) # edge 98, node 151, contrast 49
contrast_plot(118,59) # edge 118, node 141, contrast 59
contrast_plot(164,82) # edge 164, node 118, contrast 82
contrast_plot(32,16) # last edge 32, node 184, contrast 16
```



It is clear to see nodes with shift edges being detected have mean of contrasts close to zero. In some cases when those shift edges have not been detected have very different contrast values. Most of contrasts with shift edge not being detected have larger variances than variance of contrasts with shift edges being detected. Edge 77 is especially strange because it does not follow last rule. (Need some explanation)

```
load("unknownconfig_alpha_4.2.RData")
load("unknownconfig_shifts_4.2.RData")
load("unknownconfig_sigma2_4.2.RData")
load("unknownconfig_mu_4.2.RData")
load("unknownconfig_shiftvalues_4.2.RData")
load("unknownconfig_contrast_4.2.RData")
#Calculate mean of contrasts
contrast_mean5=colMeans(contrast_table5)
head(contrast_mean5)

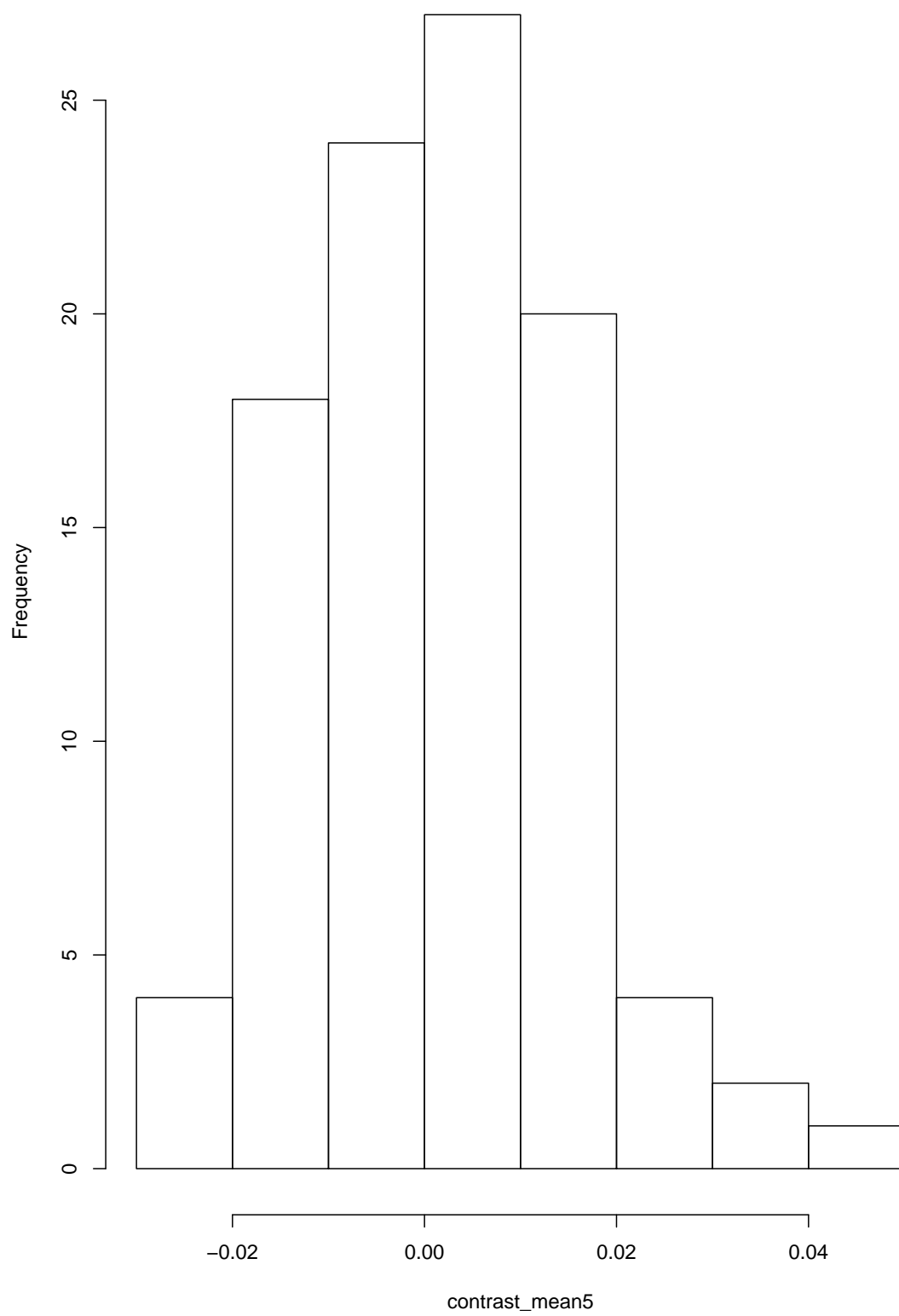
## [1] -0.013177648  0.004979592 -0.002567778 -0.013514359  0.020714921
## [6]  0.015617662

max(abs(contrast_mean5))

## [1] 0.04368802

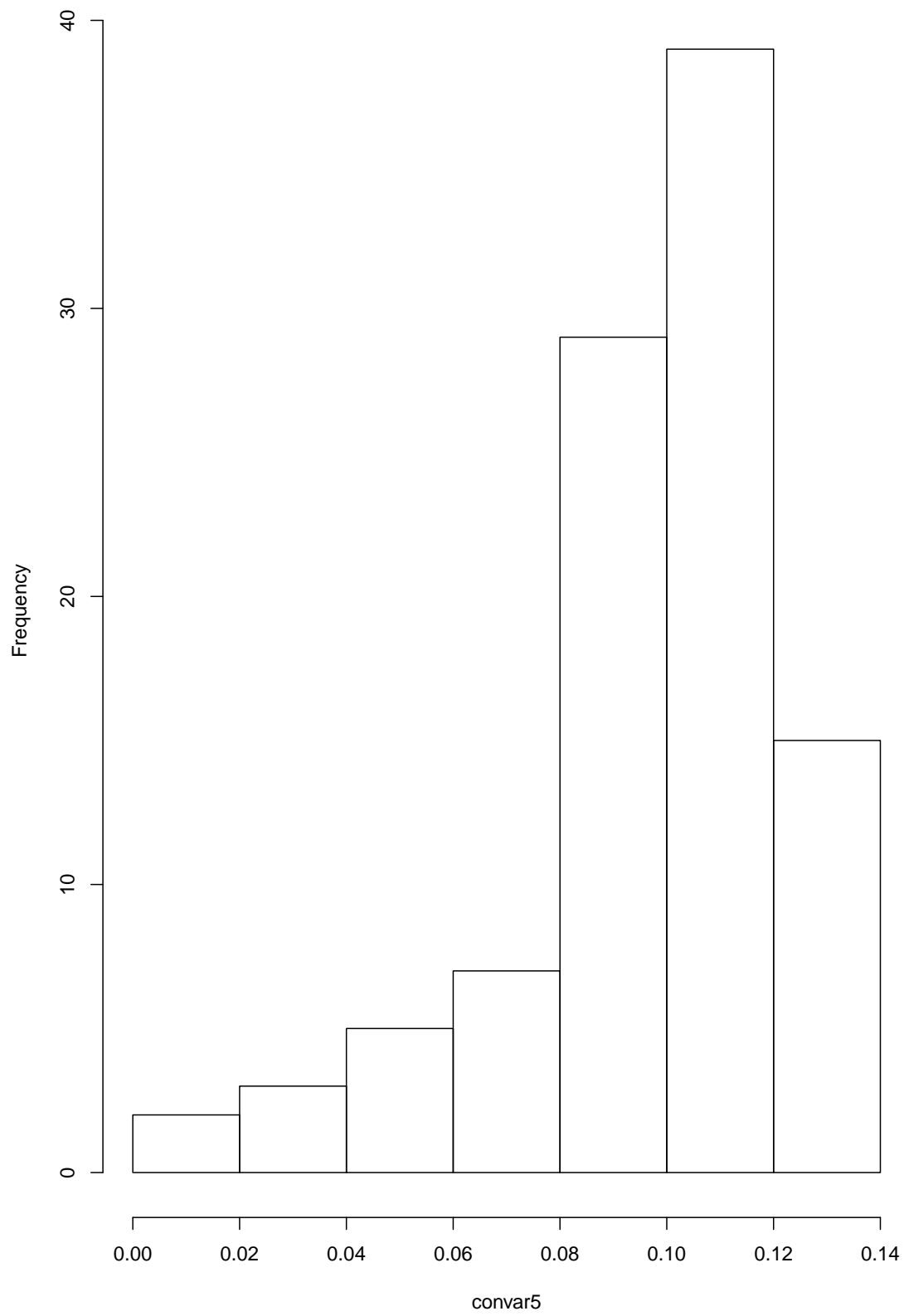
hist(contrast_mean5)
```

Histogram of contrast_mean5



```
#Calculate variance of contrasts  
convar5=apply(contrast_table5,2,ss)  
hist(convar5)
```

Histogram of convar5



#Shifts that occur the most often

```
vectorOfShift_2=unlist(vectorOfShift2, recursive = TRUE, use.names = F)
freq_table2=as.data.frame(table(vectorOfShift_2))
freq_table2
```

##	vectorOfShift_2	Freq
## 1	1	1
## 2	2	1
## 3	3	15
## 4	4	6
## 5	5	17
## 6	6	10
## 7	7	2
## 8	8	2
## 9	9	5
## 10	10	12
## 11	11	14
## 12	12	21
## 13	13	20
## 14	14	777
## 15	15	30
## 16	16	31
## 17	25	34
## 18	26	24
## 19	27	13
## 20	28	12
## 21	29	32
## 22	30	28
## 23	31	55
## 24	32	782
## 25	33	24
## 26	34	12
## 27	35	2
## 28	36	5
## 29	39	6
## 30	40	14
## 31	41	13
## 32	42	13
## 33	43	19
## 34	44	16
## 35	47	6
## 36	48	16
## 37	49	1
## 38	50	5
## 39	51	6
## 40	52	14
## 41	53	2
## 42	54	1
## 43	55	789
## 44	56	43
## 45	57	1
## 46	58	1
## 47	59	1
## 48	61	6

## 49	62	6
## 50	63	2
## 51	64	9
## 52	65	4
## 53	67	2
## 54	68	9
## 55	69	24
## 56	70	20
## 57	71	30
## 58	72	18
## 59	73	29
## 60	74	752
## 61	77	795
## 62	78	41
## 63	79	35
## 64	80	31
## 65	82	1
## 66	83	1
## 67	84	1
## 68	89	1
## 69	90	1
## 70	91	6
## 71	92	6
## 72	93	7
## 73	94	10
## 74	95	21
## 75	96	6
## 76	97	41
## 77	98	794
## 78	99	4
## 79	100	4
## 80	101	2
## 81	103	6
## 82	104	3
## 83	105	5
## 84	106	3
## 85	107	10
## 86	108	10
## 87	109	5
## 88	110	19
## 89	111	18
## 90	112	14
## 91	113	19
## 92	114	11
## 93	115	30
## 94	116	28
## 95	117	21
## 96	118	800
## 97	119	9
## 98	120	13
## 99	121	1
## 100	122	5
## 101	123	19
## 102	124	12

## 103	125	29
## 104	126	14
## 105	127	28
## 106	128	33
## 107	129	32
## 108	130	37
## 109	131	36
## 110	132	31
## 111	133	1
## 112	134	2
## 113	135	7
## 114	136	7
## 115	137	14
## 116	138	6
## 117	139	23
## 118	140	9
## 119	141	9
## 120	142	6
## 121	143	2
## 122	144	6
## 123	145	6
## 124	146	6
## 125	147	14
## 126	148	16
## 127	149	18
## 128	150	10
## 129	151	3
## 130	152	2
## 131	153	23
## 132	154	29
## 133	155	32
## 134	156	33
## 135	157	16
## 136	158	5
## 137	160	4
## 138	161	3
## 139	162	2
## 140	163	28
## 141	164	781
## 142	165	24
## 143	166	26
## 144	169	4
## 145	170	4
## 146	171	5
## 147	172	3
## 148	173	1
## 149	175	4
## 150	176	2
## 151	177	9
## 152	178	9
## 153	179	24
## 154	180	10
## 155	181	26
## 156	182	36

```
## 157      183      1
## 158      186      1
## 159      187     22
## 160      188     15
## 161      189     18
## 162      190     41
## 163      191     34
## 164      192     29
## 165      193     39
## 166      194     42
## 167      195     45
## 168      196     31
## 169      197     74
```

Try to find the contrast with variance relatively small, median and large.

```
ind_small5=which(convar5<0.04)
ind_median5=which(convar5<0.06&convar5>=0.04)
ind_large5=which(convar5>=0.085)
median_node5=intnode[ind_median5]
small_node5=intnode[ind_small5]
large_node5=as.numeric(intnode[ind_large5])
large_node5
```

```
## [1] 199 198 197 196 195 194 192 188 187 186 185 183 182 180 179 178 176
## [18] 175 174 173 170 169 168 167 166 165 164 162 160 157 154 153 152 150
## [35] 149 148 147 146 145 144 143 142 140 139 138 137 136 135 133 132 131
## [52] 130 129 128 127 126 125 124 123 122 121 120 119 117 115 114 112 111
## [69] 110 109 108 107 106 105 104 103
```

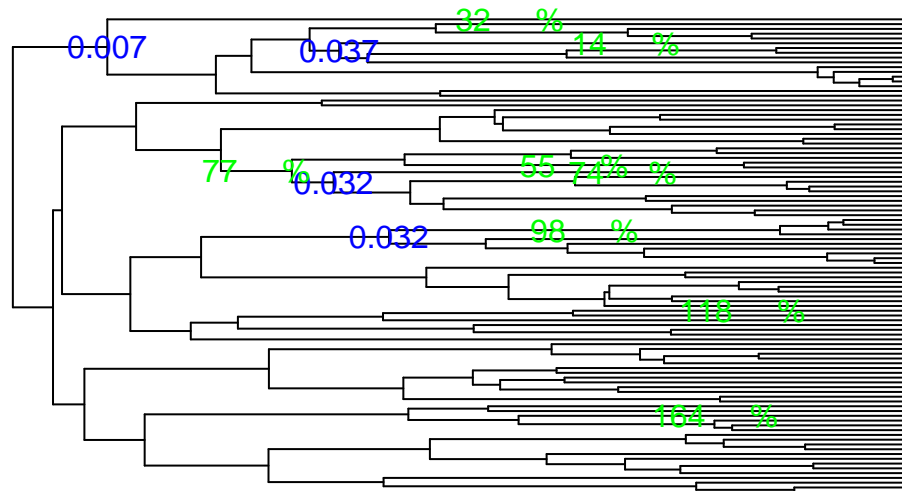
```
mean(convar5[-large_node5])
```

```
## [1] 0.09682729
```

Even though we ignore those nodes with large variance of contrasts, the variances of contrasts on average are still overestimated.

```
mean_large5=intnode[which(abs(contrast_mean5)>0.05)]
median_node5=intnode[ind_median5]
small_node5=intnode[ind_small5]
large_node5=intnode[ind_large5]
plot(eModel$tree,show.tip.label = F,main="small,median,large contrast nodes",font=8)
node.labels(round(convar5[ind_small5],3),small_node5,col="blue",frame = "none")
for(i in 1:length(shift_config)){
  edge.labels(paste(shift_config[i]," ",round(freq_table2$Freq[which(freq_table2$vectorOfShift_1==shift_c
}
})
```

small,median,large contrast nodes



#56,73

The shifts being detected most often are: 32,77,118,164,98,14,55,74 which are exactly corresponding to the shift configuration 55, 98, 118, 74, 14, 77, 32, 164 of the true model. A few nodes with shift edges have larger than average mean of contrasts. So, their variances are overestimated accordingly.

Contrasts of node with shift edges being detected or not

```
contrast_plot = function(edge, contrast){
  contrast_detect=rep(NA, 600)
  for(i in 1:length(vectorOfShift2)){
    if(edge%in%vectorOfShift2[[i]]){
      contrast_detect[i]=TRUE
    }
    else {
      contrast_detect[i]=FALSE
    }
  }
  plot(contrast_table5[,contrast] ~ factor(contrast_detect), xlab="detected?", ylab=paste(contrast, "th c
  mtext(paste("edge: ", edge, " contrast: ", contrast,
    ", detected: ", round(100*sum(contrast_detect)/800, 1), "%", sep=""))
  abline(h=0, lty=3)
  convar_bygroup = tapply(contrast_table5[,contrast], contrast_detect, var)
  mtext(side=1, at=1:2, text=round(convar_bygroup,4), line=-2)
}
```

Those figures show below represent contrasts of different nodes with group1(shift detected) and group2(shift not detected).

```
layout(matrix(1:4,2,2))
contrast_plot(14,7) # edge 14, node 193, contrast 7
contrast_plot(55,28) # edge 55, node 172, contrast 28
contrast_plot(74,37) # edge 74, node 163, contrast 37
contrast_plot(77,39) # edge 77, node 161, contrast 39

contrast_plot(98,49) # edge 98, node 151, contrast 49
contrast_plot(118,59) # edge 118, node 141, contrast 59
contrast_plot(164,82) # edge 164, node 118, contrast 82
contrast_plot(32,16) # last edge 32, node 184 , contrast 16
```