Uterine Leiomyoma Beadchip Gene Expressions

Janis Corona

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This is to re-examine the UL and non-UL samples from the Gene Expression Omnibus online data repository (GEO) for genotypes in the ULs compared to those samples without tumor tissue in them. The accession IDs for the Series is GSE95101 and for the platform is GPL13376

Lets look at some of these copy number variants of one gene with seven copy number variants or CNVs and see where the changes in the nucleotide sequences occur. Copy number variations in nucleotides can have short repeats, jumps in sequence, or deletions of a gene. I have been calling these CNVs genotypes, which are the traits and alleles responsible for the physical traits or phenotypes of an organism. Some CNVs are responsible for diseases, and in tumors there are many different CNVs that are found to be responsible. A uterine leiomyoma or fibroid is a benign tumor. These samples were taken from uterus tissue with these uternine tumors and the same neighboring uterine tissue without uterine tumors.

```
library(dplyr)
library(tidyr)
library(e1071)
library(caret)
library(randomForest)
library(MASS)
library(gbm)

UL1a <- read.csv('UL1a.csv', sep=',',
                 header=T, na.strings=c('',' '))
UL1b <- read.csv('UL1b.csv', sep=',',
                 header=T, na.strings=c('',' '))
UL1c <- read.csv('UL1c.csv', sep=',',
                 header=T, na.strings=c('',' '))
UL1d <- read.csv('UL1d.csv', sep=',',
                 header=T, na.strings=c('',' '))

UL1 <- rbind(UL1a,UL1b,UL1c,UL1d)
rm(UL1a,UL1b,UL1c,UL1d)
str(UL1)

## 'data.frame':    48701 obs. of  51 variables:
##  $ X                   : int  1 2 3 4 5 6 7 8 9 10 ...
##  $ ID                  : Factor w/ 48701 levels "ILMN_1343289",..: 1 2 3
4 5 6 7 8 9 10 ...
##  $ Species             : Factor w/ 1 level "Homo sapiens": 1 1 1 1 1 1 1
```

```
1 1 1 ...
##  $ Source               : Factor w/ 3 levels "ILMN_Controls",..: 1 1 2 1 1
2 2 2 2 2 ...
##  $ Search_Key           : Factor w/ 46721 levels
"ILMN_10001","ILMN_10014",..: 11664 11664 11664 11664 11664 11664 11664 9848
11306 1417 ...
##  $ Transcript           : Factor w/ 46724 levels
"ILMN_10001","ILMN_10014",..: 2290 2291 1377 2292 2293 4903 1218 9858 11314
1421 ...
##  $ ILMN_Gene            : Factor w/ 43186 levels "1-Dec","1-Mar",..: 8904
10174 2074 10143 10156 88 2713 4872 9250 2029 ...
##  $ Source_Reference_ID  : Factor w/ 46721 levels "NM_000015.1",..: 776
4636 1225 1123 1827 1130 1438 9379 7158 10894 ...
##  $ RefSeq_ID            : Factor w/ 28570 levels "NM_000015.1",..: 776
4636 1225 1123 1827 1130 1438 9379 7158 10894 ...
##  $ Unigene_ID           : Factor w/ 18153 levels "NA","Hs.100554",..: 1 1
1 1 1 1 1 1 1 1 ...
##  $ Entrez_Gene_ID       : Factor w/ 16063 levels "10","10000","10001",..:
10744 10744 1487 10744 10744 5871 2331 6516 10718 5545 ...
##  $ GI                   : int  14141192 20149305 25453469 4507728 4507744
5016088 7669491 88954077 33469136 89070645 ...
##  $ Accession            : Factor w/ 46721 levels "NM_000015.1",..: 776
4636 1225 1123 1827 1130 1438 9379 7158 10894 ...
##  $ Symbol               : Factor w/ 25036 levels "1-Dec","1-Mar",..: 8904
10174 2074 10143 10156 88 2713 4872 9250 2029 ...
##  $ Protein_Product      : Factor w/ 28173 levels "NA","NP_000006.1",..: 1
1 1224 1 1 1129 1437 9300 7155 10815 ...
##  $ Probe_Id             : Factor w/ 48701 levels "ILMN_1343289",..: 1 2 3
4 5 6 7 8 9 10 ...
##  $ Array_Address_Id     : int  2140735 6550370 2690379 4590356 4260048
5860528 1770601 50270 3310274 7040079 ...
##  $ Probe_Type           : Factor w/ 3 levels "A","I","S": 3 3 3 3 3 3 3 3
3 2 ...
##  $ Probe_Start          : int  416 1856 1293 1408 72 1725 930 1 1103 2975
...
##  $ SEQUENCE             : Factor w/ 48701 levels
"AAAAAACAGGAATAGCTCTAGGAGTCCTTACACAGGTCCGAGGGACCAGC",..: 9453 9026 11512 4877
6702 9532 5532 1967 10077 11476 ...
##  $ Chromosome           : Factor w/ 56 levels "1","10","11",..: 27 27 22
27 27 23 4 14 1 27 ...
##  $ Probe_Chr_Orientation: Factor w/ 3 levels "-","+","NA": 3 3 1 3 3 1 2 2
1 3 ...
##  $ Probe_Coordinates    : Factor w/ 41351 levels "100000925-100000974",..:
10161 10161 8915 10161 10161 7449 8192 4231 2830 10161 ...
##  $ Cytoband             : Factor w/ 3676 levels "10p11.1d","10p11.21a",..:
2501 2501 2039 2501 2501 2165 288 1490 1036 2033 ...
##  $ Definition           : Factor w/ 46614 levels "Homo sapiens 1-
acylglycerol-3-phosphate O-acyltransferase 1 (lysophosphatidic acid
acyltransferase, alpha) (AGP"| __truncated__,..: 5692 7144 2221 7047 6692 98
2786 8195 6195 7895 ...
```

```
##  $ Ontology_Component   : Factor w/ 7849 levels "A 20S multiprotein
assembly of total mass about 1.2 MDa that activates dynein-based activity in
vivo. A large s"| __truncated__,..: 2614 1893 1304 1321 1266 2112 1468 1893
1763 269 ...
##  $ Ontology_Process     : Factor w/ 8950 levels "[goid 6069] [pmid
1755855] [evidence IDA]; A change in state or activity of a cell or an
organism (in terms of "| __truncated__,..: 2067 2257 3716 1085 547 417 1784
1091 1091 962 ...
##  $ Ontology_Function    : Factor w/ 9453 levels "[goid 16505] [pmid
10426319] [evidence NAS]",..: 3847 2486 1975 1911 2750 2514 95 3637 3637 828
...
##  $ Synonyms             : Factor w/ 16472 levels "0610037N12Rik; RPP20;
RPP2",..: 4591 4591 5325 4591 4591 5300 2402 4591 3649 4591 ...
##  $ Obsolete_Probe_Id    : Factor w/ 16878 levels "0610037N12Rik; RPP20;
RPP2",..: 4784 4784 645 6450 1251 5508 2490 4784 3785 4784 ...
##  $ GB_ACC               : Factor w/ 46717 levels "NA","NM_000015.1",..:
777 4635 1225 1123 1827 1130 1438 9377 7156 10892 ...
##  $ GSM2496185           : num  13942 23759 27434 3092 6857 ...
##  $ GSM2496186           : num  12934 15091 26473 4269 7799 ...
##  $ GSM2496187           : num  11909 22609 23964 3455 7954 ...
##  $ GSM2496188           : num  12147 18225 27823 4258 7380 ...
##  $ GSM2496189           : num  14142 20728 24486 3333 6445 ...
##  $ GSM2496190           : num  11650 19582 26225 4545 9215 ...
##  $ GSM2496191           : num  12786 19105 28200 3413 10031 ...
##  $ GSM2496192           : num  9383 10008 27997 3191 7428 ...
##  $ GSM2496193           : num  11481 10575 23172 3597 7712 ...
##  $ GSM2496203           : num  4136 1028 16324 4994 6466 ...
##  $ GSM2496204           : num  11458 17921 26664 3095 7471 ...
##  $ GSM2496205           : num  15445 18186 25687 3138 7047 ...
##  $ GSM2496206           : num  11098 8905 22094 2473 6307 ...
##  $ GSM2496207           : num  11510 9721 21161 4353 4826 ...
##  $ GSM2496208           : num  11446 11451 26427 3863 7069 ...
##  $ GSM2496209           : num  9945 16387 27837 3027 6462 ...
##  $ GSM2496217           : num  12707 18456 28792 3251 8407 ...
##  $ GSM2496218           : num  12261 19342 25018 2322 6925 ...
##  $ GSM2496219           : num  11087 9198 27179 4554 9100 ...
##  $ GSM2496220           : num  11746 21023 29030 4131 7771 ...
```

```
nonUL1a <- read.csv('nonUL1a.csv', sep=',',
                header=T, na.strings=c('',' '))
nonUL1b <- read.csv('nonUL1b.csv', sep=',',
                header=T, na.strings=c('',' '))
nonUL1c <- read.csv('nonUL1c.csv', sep=',',
                header=T, na.strings=c('',' '))
nonUL1d <- read.csv('nonUL1d.csv', sep=',',
                header=T, na.strings=c('',' '))

nonUL1 <- rbind(nonUL1a,nonUL1b,nonUL1c,nonUL1d)
```

```
rm(nonUL1a,nonUL1b,nonUL1c,nonUL1d)
str(nonUL1)
```

```
## 'data.frame':    48701 obs. of  49 variables:
##  $ X                    : int  1 2 3 4 5 6 7 8 9 10 ...
##  $ ID                   : Factor w/ 48701 levels "ILMN_1343289",..: 1 2 3
4 5 6 7 8 9 10 ...
##  $ Species              : Factor w/ 1 level "Homo sapiens": 1 1 1 1 1 1 1
1 1 1 ...
##  $ Source               : Factor w/ 3 levels "ILMN_Controls",..: 1 1 2 1 1
2 2 2 2 2 ...
##  $ Search_Key           : Factor w/ 46721 levels
"ILMN_10001","ILMN_10014",..: 11664 11664 11664 11664 11664 11664 11664 9848
11306 1417 ...
##  $ Transcript           : Factor w/ 46724 levels
"ILMN_10001","ILMN_10014",..: 2290 2291 1377 2292 2293 4903 1218 9858 11314
1421 ...
##  $ ILMN_Gene            : Factor w/ 43186 levels "1-Dec","1-Mar",..: 8904
10174 2074 10143 10156 88 2713 4872 9250 2029 ...
##  $ Source_Reference_ID  : Factor w/ 46721 levels "NM_000015.1",..: 776
4636 1225 1123 1827 1130 1438 9379 7158 10894 ...
##  $ RefSeq_ID            : Factor w/ 28570 levels "NM_000015.1",..: 776
4636 1225 1123 1827 1130 1438 9379 7158 10894 ...
##  $ Unigene_ID           : Factor w/ 18153 levels "NA","Hs.100554",..: 1 1
1 1 1 1 1 1 1 1 ...
##  $ Entrez_Gene_ID       : Factor w/ 16063 levels "10","10000","10001",..:
10744 10744 1487 10744 10744 5871 2331 6516 10718 5545 ...
##  $ GI                   : int  14141192 20149305 25453469 4507728 4507744
5016088 7669491 88954077 33469136 89070645 ...
##  $ Accession            : Factor w/ 46721 levels "NM_000015.1",..: 776
4636 1225 1123 1827 1130 1438 9379 7158 10894 ...
##  $ Symbol               : Factor w/ 25036 levels "1-Dec","1-Mar",..: 8904
10174 2074 10143 10156 88 2713 4872 9250 2029 ...
##  $ Protein_Product      : Factor w/ 28173 levels "NA","NP_000006.1",..: 1
1 1224 1 1 1129 1437 9300 7155 10815 ...
##  $ Probe_Id             : Factor w/ 48701 levels "ILMN_1343289",..: 1 2 3
4 5 6 7 8 9 10 ...
##  $ Array_Address_Id     : int  2140735 6550370 2690379 4590356 4260048
5860528 1770601 50270 3310274 7040079 ...
##  $ Probe_Type           : Factor w/ 3 levels "A","I","S": 3 3 3 3 3 3 3 3
3 2 ...
##  $ Probe_Start          : int  416 1856 1293 1408 72 1725 930 1 1103 2975
...
##  $ SEQUENCE             : Factor w/ 48701 levels
"AAAAAACAGGAATAGCTCTAGGAGTCCTTACACAGGTCCGAGGGACCAGC",..: 9453 9026 11512 4877
6702 9532 5532 1967 10077 11476 ...
##  $ Chromosome           : Factor w/ 56 levels "1","10","11",..: 27 27 22
27 27 23 4 14 1 27 ...
##  $ Probe_Chr_Orientation: Factor w/ 3 levels "-","+","NA": 3 3 1 3 3 1 2 2
1 3 ...
```

```
##  $ Probe_Coordinates    : Factor w/ 41351 levels "100000925-100000974",..:
10161 10161 8915 10161 10161 7449 8192 4231 2830 10161 ...
##  $ Cytoband             : Factor w/ 3676 levels "10p11.1d","10p11.21a",..:
2501 2501 2039 2501 2501 2165 288 1490 1036 2033 ...
##  $ Definition           : Factor w/ 46614 levels "Homo sapiens 1-
acylglycerol-3-phosphate O-acyltransferase 1 (lysophosphatidic acid
acyltransferase, alpha) (AGP"| __truncated__,..: 5692 7144 2221 7047 6692 98
2786 8195 6195 7895 ...
##  $ Ontology_Component   : Factor w/ 7849 levels "A 20S multiprotein
assembly of total mass about 1.2 MDa that activates dynein-based activity in
vivo. A large s"| __truncated__,..: 2614 1893 1304 1321 1266 2112 1468 1893
1763 269 ...
##  $ Ontology_Process     : Factor w/ 8950 levels "[goid 6069] [pmid
1755855] [evidence IDA]; A change in state or activity of a cell or an
organism (in terms of "| __truncated__,..: 2067 2257 3716 1085 547 417 1784
1091 1091 962 ...
##  $ Ontology_Function    : Factor w/ 9453 levels "[goid 16505] [pmid
10426319] [evidence NAS]",..: 3847 2486 1975 1911 2750 2514 95 3637 3637 828
...
##  $ Synonyms             : Factor w/ 16472 levels "0610037N12Rik; RPP20;
RPP2",..: 4591 4591 5325 4591 4591 5300 2402 4591 3649 4591 ...
##  $ Obsolete_Probe_Id    : Factor w/ 16878 levels "0610037N12Rik; RPP20;
RPP2",..: 4784 4784 645 6450 1251 5508 2490 4784 3785 4784 ...
##  $ GB_ACC               : Factor w/ 46717 levels "NA","NM_000015.1",..:
777 4635 1225 1123 1827 1130 1438 9377 7156 10892 ...
##  $ GSM2496194           : num  9823 18157 27796 3428 7706 ...
##  $ GSM2496195           : num  11265 20893 24042 4279 9407 ...
##  $ GSM2496196           : num  13016 20943 24368 3049 9110 ...
##  $ GSM2496197           : num  11698 18242 24179 3574 7935 ...
##  $ GSM2496198           : num  11448 20998 25276 2178 7307 ...
##  $ GSM2496199           : num  11454 21756 26935 3768 8928 ...
##  $ GSM2496200           : num  11514 21849 26969 3170 9457 ...
##  $ GSM2496201           : num  10621 10200 24231 2292 7765 ...
##  $ GSM2496202           : num  11066 9349 22945 4513 8454 ...
##  $ GSM2496210           : num  10189 21816 29280 4816 8773 ...
##  $ GSM2496211           : num  9998 18435 26231 4683 8579 ...
##  $ GSM2496212           : num  11407 23942 27389 3589 7977 ...
##  $ GSM2496213           : num  9476 10440 23432 5444 8126 ...
##  $ GSM2496214           : num  11708 9478 22640 4533 8418 ...
##  $ GSM2496215           : num  11457 11803 24008 5839 8549 ...
##  $ GSM2496216           : num  12900 20375 28086 4044 7252 ...
##  $ GSM2496221           : num  10020 16842 25324 2469 7225 ...
##  $ GSM2496222           : num  13409 9030 22273 3315 7844 ...
```

```
UL <- UL1[,-c(1:13,15:19,21,29:31)]
nonUL <- nonUL1[,-c(1:13,15:19,21,29:31)]

write.csv(UL,'UL.csv', row.names=FALSE)
write.csv(nonUL, 'nonUL.csv', row.names=FALSE)
```

```
fibroid <- read.csv('UL.csv', sep=',', header=T, na.strings=c('',' '))
nonFibroid <- read.csv('nonUL.csv', sep=',', header=T, na.strings=c('',' '))

fibroid_gene_n <- fibroid %>% group_by(Symbol) %>% count(n())
narm <- grep('^NA$',fibroid_gene_n$Symbol)

fibroid1 <- fibroid_gene_n[-narm,-2]
colnames(fibroid1)[2] <- 'gene_count'

NONfibroid_gene_n <- nonFibroid %>% group_by(Symbol) %>% count(n())
narm1 <- grep('^NA$',NONfibroid_gene_n$Symbol)

nonFibroid1 <- NONfibroid_gene_n[-narm1,-2]
colnames(nonFibroid1)[2] <- 'gene_count'

GeneCopyNumberVariants <-
fibroid1[order(fibroid1$gene_count,decreasing=TRUE)[1:10],]
GeneCopyNumberVariants

## # A tibble: 10 x 2
## # Groups:   Symbol [10]
##    Symbol    gene_count
##    <fct>          <int>
##  1 DDX12             10
##  2 KIAA0692           9
##  3 LOC23117           8
##  4 PLEC1              8
##  5 BDNF               7
##  6 CTNNB1             7
##  7 DMD                7
##  8 LOC202134          7
##  9 LOC339047          7
## 10 LOC653086          7
```

Combine the gene counts with the tables of samples for each type of UL or nonUL.

```
Fibroid_count <- merge(fibroid1, fibroid, by.x='Symbol', by.y='Symbol')
nonFibroid_count <- merge(nonFibroid1, nonFibroid, by.x='Symbol',
by.y='Symbol')
Fibroid_count[order(Fibroid_count$gene_count, decreasing=TRUE)[1:20],1:3]

##         Symbol gene_count
SEQUENCE
## 5646     DDX12         10
CCAGTCCCTGACTACAGAGGATTTCCCCAAAGTCCCTGGCTGTGAGGTTC
## 5647     DDX12         10
TTACTGGGGATGGTATTTAGGAGCCAGGAAAGCCGGTGCATTCCTAGTGA
## 5648     DDX12         10
TCTCCTGCCCCCTCCGGAAGCTTGGATGCCCCTCCACACCCTCTTGATCT
## 5649     DDX12         10
CAGACTTCTCGCTTCCTTTCTGCTGGGCCTCTGAGGGGTCATGGGGCCAT
```

```
## 5650     DDX12         10
ACATGTGCTGTCACTGGAACTTGCTCTTTTCACTCAGCAGCCAGAGGGTC
## 5651     DDX12         10
AAACGTTACAGTGTTCCGATGAGACACAGTAGGCAGTACTTGGGAGGGTC
## 5652     DDX12         10
CAGGGCAGGAACCACGTCTTTACAGTTTGATGTTCCCAGAGCTGACCCAG
## 5653     DDX12         10
GCAGGGGAGATTGGGTTTAGGGGCTTTCCTGGTCTGCATTCTGCTACAGC
## 5654     DDX12         10
CCGCCGGGCTGCTTTTTCCTTGGATGCCCATCAGGACGCCTCAGTTCTCT
## 5655     DDX12         10
CGTTGCTACAAGCTGTTTTTTGAATGTCTCTACACAGTCCAGGCAGGAAG
## 10983 KIAA0692          9
AAGTGGTGCCTGGCTGTCCCTATACTGTGCTGCTGGGTGTTCCAGCCTGT
## 10984 KIAA0692          9
TAAGTGCAGTGAGCTCTGGCGGAAACCACCCTCTGCCCCGTCTGTTGGAT
## 10985 KIAA0692          9
CATTGTAATGATAAGGAAATGTTGCGATCAAATAAGATTTAGACACACTT
## 10986 KIAA0692          9
GATCACAGGCACAGGGAAGCCACAAGGAGCTCTGTATGAGTTGTGTTTGC
## 10987 KIAA0692          9
CAGGCGACTGGGTAGCAGATGTGGAAGCTGATGGTTAGGCCCAGGGCATG
## 10988 KIAA0692          9
GTTGTTCTGGACGATCTTCGGGATCCTCTGGGGCACTGTGACACTCGGAG
## 10989 KIAA0692          9
GAGTGCTGGGAAGGTTAATGTTAAATGGGTTGTGTGTCGGGGAGGGTACA
## 10990 KIAA0692          9
AGCTCCACCTTGACCCAGCCTCACAACAAAAAGTTTGTGTATGACCAGGC
## 10991 KIAA0692          9
GCAAATGTAACTCAGGGGTTTGGGGCCAGAGGAAGAGGGAGAAGGTGGCC
## 12174 LOC23117          8
CTGGCCTTCCCTCATCAGCCGTAAATGATGATTTACTGCTGTTACCATCA
```

Add a mean, median, min, and max column to these tables.

```
Fibroid_count$Fibroid_Mean <- rowMeans(Fibroid_count[11:30])
nonFibroid_count$nonFibroid_Mean <- rowMeans(nonFibroid_count[11:28])
```

Use the tidyr package to group by sample ID by gathering those columns into one.

```
UL_3 <- gather(Fibroid_count, 'UL_Sample_ID','Value',11:30)
nonUL_3 <- gather(nonFibroid_count, 'nonUL_Sample_ID', 'Value',11:28)
```

Create the stat tables then combine for the UL and nonUL sample sets using the dplyr package.

```
UL_median <- UL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value),
median)
colnames(UL_median)[2] <- 'Fibroid_Median'

nonUL_median <- nonUL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value),
```

```
median)
colnames(nonUL_median)[2] <- 'nonFibroid_Median'

UL_max <- UL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value), max)
colnames(UL_max)[2] <- 'Fibroid_max'

nonUL_max <- nonUL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value),
max)
colnames(nonUL_max)[2] <- 'nonFibroid_max'

UL_min <- UL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value), min)
colnames(UL_min)[2] <- 'Fibroid_min'

nonUL_min <- nonUL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value),
min)
colnames(nonUL_min)[2] <- 'nonFibroid_min'

UL_sd <- UL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value), sd)
colnames(UL_sd)[2] <- 'Fibroid_stdError'

nonUL_sd <- nonUL_3 %>% group_by(SEQUENCE) %>% summarise_at(vars(Value), sd)
colnames(nonUL_sd)[2] <- 'nonFibroid_stdError'
```

Combine these four tables together.

```
Fibroid_stats <- merge(UL_median, UL_max, by.x='SEQUENCE', by.y='SEQUENCE')
Fibroid_stats1 <- merge(Fibroid_stats, UL_min, by.x='SEQUENCE',
by.y='SEQUENCE')
Fibroid_stats2 <- merge(Fibroid_count, Fibroid_stats1, by.x='SEQUENCE',
by.y='SEQUENCE')
Fibroid_stats3 <- merge(Fibroid_stats2, UL_sd, by.x='SEQUENCE',
by.y='SEQUENCE')
colnames(Fibroid_stats3)[11:30] <- paste('UL_',
colnames(Fibroid_stats3)[11:30], sep='')

nonFibroid_stats <- merge(nonUL_median, nonUL_max, by.x='SEQUENCE',
by.y='SEQUENCE')
nonFibroid_stats1 <- merge(nonFibroid_stats, nonUL_min, by.x='SEQUENCE',
by.y='SEQUENCE')
nonFibroid_stats2 <- merge(nonFibroid_count, nonFibroid_stats1,
by.x='SEQUENCE', by.y='SEQUENCE')
nonFibroid_stats3 <- merge(nonFibroid_stats2, nonUL_sd, by.x='SEQUENCE',
by.y='SEQUENCE')
colnames(nonFibroid_stats3)[11:28] <- paste('nonUL_',
colnames(nonFibroid_stats3)[11:28], sep='')


nonfibroid <- nonFibroid_stats3[,c(1,11:33)]
```

```
all <- merge(Fibroid_stats3, nonfibroid, by.x='SEQUENCE', by.y='SEQUENCE')
str(all)
```

```
## 'data.frame':    30549 obs. of  58 variables:
##  $ SEQUENCE             : Factor w/ 48701 levels
"AAAAAACAAAACCGCGCAGCGGAGAACCGGTGCCTGAGTCTCCCAGGGAC",..: 1 4 5 7 8 10 12 13
15 18 ...
##  $ Symbol               : Factor w/ 25036 levels "1-Dec","1-Mar",..: 12031
11383 13002 14397 15611 12721 12474 10953 24822 18000 ...
##  $ gene_count           : int  1 1 1 1 1 1 1 1 1 1 ...
##  $ Probe_Chr_Orientation: Factor w/ 3 levels "-","+","NA": 2 1 2 3 3 2 2 3
1 2 ...
##  $ Probe_Coordinates    : Factor w/ 41351 levels "100000925-100000974",..:
33600 3552 19430 41351 41351 36542 28872 41351 31503 41119 ...
##  $ Cytoband             : Factor w/ 3676 levels "10p11.1d","10p11.21a",..:
3472 3472 3472 3472 3472 293 3472 3472 1261 2180 ...
##  $ Definition           : Factor w/ 46614 levels "{3 region, probe S2}
[human, 76N, mammary epithelial cells, mRNA Partial, 339 nt]",..: 33743 33388
39229 37744 38664 34103 33968 35730 29449 21454 ...
##  $ Ontology_Component   : Factor w/ 7849 levels "A 20S multiprotein
assembly of total mass about 1.2 MDa that activates dynein-based activity in
vivo. A large s"| __truncated__,..: 4150 4150 4150 4150 4150 4150 4150 4150
5434 6267 ...
##  $ Ontology_Process     : Factor w/ 8950 levels "[goid 19642] [evidence
IEA]; The chemical reactions and pathways involving carbohydrates, any of a
group of org"| __truncated__,..: 2492 2492 2492 2492 2492 2492 2492 2492 8502
3316 ...
##  $ Ontology_Function    : Factor w/ 9453 levels "[goid 15280] [evidence
IEA]; Interacting selectively with sodium ions (Na+) [goid 31402] [evidence
IEA]",..: 8111 8111 8111 8111 8111 8111 8111 8111 7071 2705 ...
##  $ UL_GSM2496185        : num  49.5 51.2 59.9 312.3 52.3 ...
##  $ UL_GSM2496186        : num  51.5 52.3 64.5 333.3 53.7 ...
##  $ UL_GSM2496187        : num  46.9 49 50.3 331.8 56.4 ...
##  $ UL_GSM2496188        : num  50.1 48.4 52 360.9 51.8 ...
##  $ UL_GSM2496189        : num  54 49.8 52.8 339 52.6 ...
##  $ UL_GSM2496190        : num  50.1 48.6 52 411.7 51.9 ...
##  $ UL_GSM2496191        : num  49.5 46 54.7 424.3 54.8 ...
##  $ UL_GSM2496192        : num  46.5 50.6 50.2 517.3 53.6 ...
##  $ UL_GSM2496193        : num  48 46.7 52.9 631.7 54.1 ...
##  $ UL_GSM2496203        : num  48.1 51.5 55.1 576.7 55 ...
##  $ UL_GSM2496204        : num  51.7 50.8 51.6 300.4 52.3 ...
##  $ UL_GSM2496205        : num  46.9 47.5 52.3 348.9 53.2 ...
##  $ UL_GSM2496206        : num  50.5 43.9 54.9 722.6 56.1 ...
##  $ UL_GSM2496207        : num  44 46.5 56.5 1218 55.9 ...
##  $ UL_GSM2496208        : num  51.1 46 51.9 535.5 55.5 ...
##  $ UL_GSM2496209        : num  55.2 46.9 50.6 669 53.3 ...
##  $ UL_GSM2496217        : num  48.9 45.8 49.6 361.9 49.8 ...
##  $ UL_GSM2496218        : num  50 49.9 53.9 377.2 53.6 ...
##  $ UL_GSM2496219        : num  48.3 49.5 50.4 544.1 58.1 ...
##  $ UL_GSM2496220        : num  47.7 46.7 51.7 406.1 56.3 ...
```

```
##  $ Fibroid_Mean         : num  49.4 48.4 53.4 486.1 54 ...
##  $ Fibroid_Median       : num  49.5 48.5 52.1 408.9 53.7 ...
##  $ Fibroid_max          : num  55.2 52.3 64.5 1218 58.1 ...
##  $ Fibroid_min          : num  44 43.9 49.6 300.4 49.8 ...
##  $ Fibroid_stdError     : num  2.59 2.3 3.61 214.26 2 ...
##  $ nonUL_GSM2496194     : num  42.7 42.7 56.6 442 57 ...
##  $ nonUL_GSM2496195     : num  49.1 45.3 56 505.4 55.3 ...
##  $ nonUL_GSM2496196     : num  51.1 45.3 55.1 416.5 57.4 ...
##  $ nonUL_GSM2496197     : num  46 52.6 56.2 475.3 54.6 ...
##  $ nonUL_GSM2496198     : num  49.1 54.1 53.7 408.1 50.8 ...
##  $ nonUL_GSM2496199     : num  52.6 47.9 51.1 510.7 52.5 ...
##  $ nonUL_GSM2496200     : num  46 47.1 52.5 681.9 50.3 ...
##  $ nonUL_GSM2496201     : num  45.9 46.8 48.3 689.3 56.7 ...
##  $ nonUL_GSM2496202     : num  50.6 45.3 56.5 735.2 48.7 ...
##  $ nonUL_GSM2496210     : num  48.3 49.3 56.3 426.9 52.5 ...
##  $ nonUL_GSM2496211     : num  46.8 46 54.6 567.3 47.4 ...
##  $ nonUL_GSM2496212     : num  50.6 48.7 54.3 493.8 51.5 ...
##  $ nonUL_GSM2496213     : num  49.1 49.5 56.7 792.8 48.8 ...
##  $ nonUL_GSM2496214     : num  48.8 49.9 54 710.5 50.6 ...
##  $ nonUL_GSM2496215     : num  48 47.1 52.9 795.4 53.1 ...
##  $ nonUL_GSM2496216     : num  47.9 49.2 48 574.5 51.1 ...
##  $ nonUL_GSM2496221     : num  48.8 48.2 50.6 507.4 54.7 ...
##  $ nonUL_GSM2496222     : num  44.6 49.5 50.4 590 56 ...
##  $ nonFibroid_Mean      : num  48.1 48 53.5 573.5 52.7 ...
##  $ nonFibroid_Median    : num  48.5 48.1 54.1 539 52.5 ...
##  $ nonFibroid_max       : num  52.6 54.1 56.7 795.4 57.4 ...
##  $ nonFibroid_min       : num  42.7 42.7 48 408.1 47.4 ...
##  $ nonFibroid_stdError  : num  2.46 2.75 2.85 130.03 3.07 ...
```

Lets change the 'fibroid' in the column names to 'UL' for uterine leiomyoma.

```
colnames(all) <- gsub('Fibroid', 'UL', colnames(all))
```

Reorder the table so that the stats are at the end of the columns.

```
All <- all[,c(1:10,11:30, 36:53,31:35,54:58)]
str(All)
```

```
## 'data.frame':    30549 obs. of  58 variables:
##  $ SEQUENCE             : Factor w/ 48701 levels
"AAAAAACAAAACCGCGCAGCGGAGAACCGGTGCCTGAGTCTCCCAGGGAC",..: 1 4 5 7 8 10 12 13
15 18 ...
##  $ Symbol               : Factor w/ 25036 levels "1-Dec","1-Mar",..: 12031
11383 13002 14397 15611 12721 12474 10953 24822 18000 ...
##  $ gene_count           : int  1 1 1 1 1 1 1 1 1 1 ...
##  $ Probe_Chr_Orientation: Factor w/ 3 levels "-","+","NA": 2 1 2 3 3 2 2 3
1 2 ...
##  $ Probe_Coordinates    : Factor w/ 41351 levels "100000925-100000974",..:
33600 3552 19430 41351 41351 36542 28872 41351 31503 41119 ...
##  $ Cytoband             : Factor w/ 3676 levels "10p11.1d","10p11.21a",..:
3472 3472 3472 3472 3472 293 3472 3472 1261 2180 ...
```

```
##  $ Definition           : Factor w/ 46614 levels "{3 region, probe S2}
[human, 76N, mammary epithelial cells, mRNA Partial, 339 nt]",..: 33743 33388
39229 37744 38664 34103 33968 35730 29449 21454 ...
##  $ Ontology_Component   : Factor w/ 7849 levels "A 20S multiprotein
assembly of total mass about 1.2 MDa that activates dynein-based activity in
vivo. A large s"| __truncated__,..: 4150 4150 4150 4150 4150 4150 4150 4150
5434 6267 ...
##  $ Ontology_Process     : Factor w/ 8950 levels "[goid 19642] [evidence
IEA]; The chemical reactions and pathways involving carbohydrates, any of a
group of org"| __truncated__,..: 2492 2492 2492 2492 2492 2492 2492 2492 8502
3316 ...
##  $ Ontology_Function    : Factor w/ 9453 levels "[goid 15280] [evidence
IEA]; Interacting selectively with sodium ions (Na+) [goid 31402] [evidence
IEA]",..: 8111 8111 8111 8111 8111 8111 8111 8111 7071 2705 ...
##  $ UL_GSM2496185        : num  49.5 51.2 59.9 312.3 52.3 ...
##  $ UL_GSM2496186        : num  51.5 52.3 64.5 333.3 53.7 ...
##  $ UL_GSM2496187        : num  46.9 49 50.3 331.8 56.4 ...
##  $ UL_GSM2496188        : num  50.1 48.4 52 360.9 51.8 ...
##  $ UL_GSM2496189        : num  54 49.8 52.8 339 52.6 ...
##  $ UL_GSM2496190        : num  50.1 48.6 52 411.7 51.9 ...
##  $ UL_GSM2496191        : num  49.5 46 54.7 424.3 54.8 ...
##  $ UL_GSM2496192        : num  46.5 50.6 50.2 517.3 53.6 ...
##  $ UL_GSM2496193        : num  48 46.7 52.9 631.7 54.1 ...
##  $ UL_GSM2496203        : num  48.1 51.5 55.1 576.7 55 ...
##  $ UL_GSM2496204        : num  51.7 50.8 51.6 300.4 52.3 ...
##  $ UL_GSM2496205        : num  46.9 47.5 52.3 348.9 53.2 ...
##  $ UL_GSM2496206        : num  50.5 43.9 54.9 722.6 56.1 ...
##  $ UL_GSM2496207        : num  44 46.5 56.5 1218 55.9 ...
##  $ UL_GSM2496208        : num  51.1 46 51.9 535.5 55.5 ...
##  $ UL_GSM2496209        : num  55.2 46.9 50.6 669 53.3 ...
##  $ UL_GSM2496217        : num  48.9 45.8 49.6 361.9 49.8 ...
##  $ UL_GSM2496218        : num  50 49.9 53.9 377.2 53.6 ...
##  $ UL_GSM2496219        : num  48.3 49.5 50.4 544.1 58.1 ...
##  $ UL_GSM2496220        : num  47.7 46.7 51.7 406.1 56.3 ...
##  $ nonUL_GSM2496194     : num  42.7 42.7 56.6 442 57 ...
##  $ nonUL_GSM2496195     : num  49.1 45.3 56 505.4 55.3 ...
##  $ nonUL_GSM2496196     : num  51.1 45.3 55.1 416.5 57.4 ...
##  $ nonUL_GSM2496197     : num  46 52.6 56.2 475.3 54.6 ...
##  $ nonUL_GSM2496198     : num  49.1 54.1 53.7 408.1 50.8 ...
##  $ nonUL_GSM2496199     : num  52.6 47.9 51.1 510.7 52.5 ...
##  $ nonUL_GSM2496200     : num  46 47.1 52.5 681.9 50.3 ...
##  $ nonUL_GSM2496201     : num  45.9 46.8 48.3 689.3 56.7 ...
##  $ nonUL_GSM2496202     : num  50.6 45.3 56.5 735.2 48.7 ...
##  $ nonUL_GSM2496210     : num  48.3 49.3 56.3 426.9 52.5 ...
##  $ nonUL_GSM2496211     : num  46.8 46 54.6 567.3 47.4 ...
##  $ nonUL_GSM2496212     : num  50.6 48.7 54.3 493.8 51.5 ...
##  $ nonUL_GSM2496213     : num  49.1 49.5 56.7 792.8 48.8 ...
##  $ nonUL_GSM2496214     : num  48.8 49.9 54 710.5 50.6 ...
##  $ nonUL_GSM2496215     : num  48 47.1 52.9 795.4 53.1 ...
##  $ nonUL_GSM2496216     : num  47.9 49.2 48 574.5 51.1 ...
```

```
##  $ nonUL_GSM2496221     : num  48.8 48.2 50.6 507.4 54.7 ...
##  $ nonUL_GSM2496222     : num  44.6 49.5 50.4 590 56 ...
##  $ UL_Mean              : num  49.4 48.4 53.4 486.1 54 ...
##  $ UL_Median            : num  49.5 48.5 52.1 408.9 53.7 ...
##  $ UL_max               : num  55.2 52.3 64.5 1218 58.1 ...
##  $ UL_min               : num  44 43.9 49.6 300.4 49.8 ...
##  $ UL_stdError          : num  2.59 2.3 3.61 214.26 2 ...
##  $ nonUL_Mean           : num  48.1 48 53.5 573.5 52.7 ...
##  $ nonUL_Median         : num  48.5 48.1 54.1 539 52.5 ...
##  $ nonUL_max            : num  52.6 54.1 56.7 795.4 57.4 ...
##  $ nonUL_min            : num  42.7 42.7 48 408.1 47.4 ...
##  $ nonUL_stdError       : num  2.46 2.75 2.85 130.03 3.07 ...

All_stats_only <- All[,c(1,2,3,49:58)]
stats_all <- All_stats_only[!duplicated(All_stats_only),]

stats_all$foldChangeMean_UL_to_nonUL <-
stats_all$UL_Mean/stats_all$nonUL_Mean

FoldChangeGenes <- stats_all[order(stats_all$foldChangeMean_UL_to_nonUL,
decreasing=TRUE)[c(1:5,30545:30549)],]

FoldChangeGenes

##                                                SEQUENCE   Symbol
gene_count
## 16709 GCAACGCTCCTCTGAAATGCTTGTCTTTTTTCTGTTGCCGAAATAGCTGG KIAA1199
1
## 17948 GCCCCAGCAAGCCTCCCTCCATCCTCCAGTGGGAAACTGTTGATGGTGTT     PENK
1
## 22790 GGTATTGCTGATCGTATGCAGAAGGAAATCACTGCTCTGGCTCCTAGCAC     ACTC
1
## 28162 TGCGAGACCTGGGTGTCCAACCTGCGCTACAACCACATGCTGCGGAAGAA     DLK1
2
## 3569  AGGCCCTGGAGGCTGCAACATACCTCAATCCTGTCCCAGGCCGGATCCTC    MMP11
1
## 17565 GCCAACCTCCTCTCACAGCCTCTGTATCTCTGCAGGCCATACTGGTTCCA    ABCA8
1
## 6582  CAGATGTTTTCCCTTGTGGCAGTCTTCAGCCTCCTCTACCCTACATGATC    ADH1A
1
## 8958  CCCAGTGACACTTCAGAGAGCTGGTAGTTAGTAGCATGTTGAGCCAGGCC      FOS
1
## 27542 TGACTGTCCCTGCCAATGCTCCAGCTGTCGTCTGACTCTGGGTTCGTTGG     FOSB
1
## 6881  CAGCTGGGCGATGTGCGAGCTGATAGTGAGCGGCAGAATCAGGAGTACCA    KRT19
1
##         UL_Mean UL_Median     UL_max   UL_min UL_stdError nonUL_Mean
## 16709 3029.4710 2566.7096  6548.8160 156.8375  1948.88193  167.75283
## 17948 1798.4359  237.7790 26134.7105  48.2000  5783.33114  120.28719
## 22790 4693.5182 4880.4535 13992.2647 110.2923  3328.78611  392.30767
```

```
## 28162  946.4365  249.4440  5799.7170  54.7250  1542.06289   87.41913
## 3569  4767.6862 3652.1430 15286.3475 235.9674  4029.71487  442.19991
## 17565  143.2650   99.0957   628.2257  56.0500   126.17477  724.33959
## 6582   608.7258  545.4995  1947.6415  54.3500   476.16852 3102.90066
## 8958  1287.1220  805.1551  7335.1691 220.6810  1573.12379 6916.73077
## 27542  205.9075   70.8273  2290.1261  53.1000   493.97933 1142.67872
## 6881   130.4870   85.6123   311.2020  45.6000    90.60451  815.61118
##       nonUL_Median  nonUL_max nonUL_min nonUL_stdError
## 16709    116.83410   514.9222   63.1000      127.90943
## 17948     71.88335   431.7050   54.2000       98.67055
## 22790    332.26735   830.2335  169.4130      197.47324
## 28162     52.70835   669.4027   47.3000      145.29680
## 3569     308.24915  1683.5592   75.9750      430.96362
## 17565    643.88430  1493.2380  125.9636      317.38002
## 6582    2961.70150  7727.8238  275.2883     1833.54639
## 8958    6722.92010 17362.0317 2406.0706     3676.04993
## 27542    595.09105  7604.0221   92.4250     1864.52510
## 6881     738.20585  2049.6529   57.4000      498.12489
##       foldChangeMean_UL_to_nonUL
## 16709                 18.0591346
## 17948                 14.9511840
## 22790                 11.9638706
## 28162                 10.8264232
## 3569                  10.7817440
## 17565                  0.1977871
## 6582                   0.1961796
## 8958                   0.1860882
## 27542                  0.1801972
## 6881                   0.1599868
```

```
str(stats_all)
```

```
## 'data.frame':    30549 obs. of  14 variables:
##  $ SEQUENCE                  : Factor w/ 48701 levels
"AAAAAACAAAACCGCGCAGCGGAGAACCGGTGCCTGAGTCTCCCAGGGAC",..: 1 4 5 7 8 10 12 13
15 18 ...
##  $ Symbol                    : Factor w/ 25036 levels "1-Dec","1-Mar",..:
12031 11383 13002 14397 15611 12721 12474 10953 24822 18000 ...
##  $ gene_count                : int  1 1 1 1 1 1 1 1 1 1 ...
##  $ UL_Mean                   : num  49.4 48.4 53.4 486.1 54 ...
##  $ UL_Median                 : num  49.5 48.5 52.1 408.9 53.7 ...
##  $ UL_max                    : num  55.2 52.3 64.5 1218 58.1 ...
##  $ UL_min                    : num  44 43.9 49.6 300.4 49.8 ...
##  $ UL_stdError               : num  2.59 2.3 3.61 214.26 2 ...
##  $ nonUL_Mean                : num  48.1 48 53.5 573.5 52.7 ...
##  $ nonUL_Median              : num  48.5 48.1 54.1 539 52.5 ...
##  $ nonUL_max                 : num  52.6 54.1 56.7 795.4 57.4 ...
##  $ nonUL_min                 : num  42.7 42.7 48 408.1 47.4 ...
##  $ nonUL_stdError            : num  2.46 2.75 2.85 130.03 3.07 ...
##  $ foldChangeMean_UL_to_nonUL: num  1.027 1.007 0.997 0.848 1.024 ...
```

```
write.csv(stats_all, 'stats_only_UL_nonUL.csv', row.names=FALSE)
```

Combine the table of top and bottom five genes in fold change values of the ratio of UL to non-UL sample means, FoldChangeGenes, with the table of the ten genes having the highest number of copy number variations or genotypes, GeneCopyNumberVariants.

```
ontology <- nonFibroid[,c(1,6:9)]
gnc <- as.data.frame(GeneCopyNumberVariants)[1]

keyGenes1 <- merge(gnc, stats_all, by.x='Symbol', by.y='Symbol')

keyGenes1a <- merge(keyGenes1, ontology, by.x='Symbol', by.y='Symbol')

keyGenes2 <- merge(FoldChangeGenes, ontology, by.x='Symbol', by.y='Symbol')
keyGenes2a <- keyGenes2[,c(1:3,15:18,4:14)]
keyGenes1b <- keyGenes1a[,c(1:3,15:18,4:14)]

KeyGenes <- rbind(keyGenes2a, keyGenes1b)
KG <- KeyGenes[!duplicated(KeyGenes$SEQUENCE),]
KG1 <- KG[order(KG$foldChangeMean_UL_to_nonUL, decreasing=TRUE),]
KG1[,c(1:3,18)]
```

```
##        Symbol                                                SEQUENCE
gene_count
## 8    KIAA1199 GCAACGCTCCTCTGAAATGCTTGTCTTTTTTCTGTTGCCGAAATAGCTGG
1
## 11       PENK GCCCCAGCAAGCCTCCCTCCATCCTCCAGTGGGAAACTGTTGATGGTGTT
1
## 2        ACTC GGTATTGCTGATCGTATGCAGAAGGAAATCACTGCTCTGGCTCCTAGCAC
1
## 4        DLK1 TGCGAGACCTGGGTGTCCAACCTGCGCTACAACCACATGCTGCGGAAGAA
2
## 10      MMP11 AGGCCCTGGAGGCTGCAACATACCTCAATCCTGTCCCAGGCCGGATCCTC
1
## 68     CTNNB1 AGCTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAG
7
## 89     CTNNB1 CTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAGCT
7
## 82     CTNNB1 AGTCTCTCGTAGTGTTAAGTTATAGTGAATACTGCTACAGCAATTTCTAA
7
## 231       DMD CAGTGTTGGGATCACTCACTTTCCCCCTACAGGACTCAGATCTGGGAGGC
7
## 210       DMD CTCCTCTCAGCTGAACACCCTCCTTTCACTCCCAAATGCAAACAGTCTCT
7
## 96     CTNNB1 GCCTCTTGCACTCTGAATTGGGAATGTTTGCACCACAGTGGGGGGCTTGC
7
## 140     DDX12 CCGCCGGGCTGCTTTTTCCTTGGATGCCCATCAGGACGCCTCAGTTCTCT
10
## 397  LOC23117 TCAACCACATCCTTCAAAAGGACTATGCCTGTTTATAAGCCCAGCTGTTT
8
```

```
## 361 LOC202134 GCCAAAGGAATGGGCTCCAGACACCCCCTCTTCCAGAGCAAGGATGAAGG
7
## 286  KIAA0692 TAAGTGCAGTGAGCTCTGGCGGAAACCACCCTCTGCCCCGTCTGTTGGAT
9
## 61     CTNNB1 CAAACTTTACAGAGGAGAATGCCCTGTTTGTTAACCATGTTTCTTTTGGC
7
## 516 LOC653086 TGATGTGTCACGCCACTGTACTCCAGCCTGACGGCAGAGCGAGACTCCAT
7
## 33       BDNF CCCTCCACCTCCTGCTCGGGGGGCTTTAATGAGACACCCACCGCTGCTGT
7
## 551     PLEC1 CCCGACGAGCAGGACTTCATCCAGGCCTACGAGGAGGTGCGCGAGAAGTA
8
## 445  LOC23117 TGTCGTTTCCTCCATTCTTCACCAAAACATCAGCGTACATAGGCACATGG
8
## 474 LOC339047 ACTGCCTGTGTGGCTCCTTGAGTGCGCGGAGGCCAAAGCTGAGATGACTT
7
## 331  KIAA0692 CATTGTAATGATAAGGAAATGTTGCGATCAAATAAGATTTAGACACACTT
9
## 559     PLEC1 CCCTCGGGCAGCCTGTTTCCCTCCCTGGTGGTTGTGGGTCACGTTGTCAC
8
## 530 LOC653086 GGTGTGCTCTGGTATGTAATGACAATATGTGAACAAACCTGTGGAATTAA
7
## 259  KIAA0692 GAGTGCTGGGAAGGTTAATGTTAAATGGGTTGTGTGTCGGGGAGGGTACA
9
## 429  LOC23117 GGCTCCTCTTTGGGCTCCTACTGGAATTTATCAGCCATCAGTGCATCTCT
8
## 252       DMD TCTATCAACAGAGCTGAATGAGTGCCAGGAAGCTGCGAAATCTGTCTTAC
7
## 268  KIAA0692 GCAAATGTAACTCAGGGGTTTGGGGCCAGAGGAAGAGGGAGAAGGTGGCC
9
## 19       BDNF AATAATAGAGTGTGGGAGTTTTGGGGCCGAAGTCTTTCCCGGAGCAGCTG
7
## 340 LOC202134 CAAACCCTTGAAGACATTTCAGGGCCATGCTCACTTGGGAGGGTTTGAGG
7
## 405  LOC23117 AAAGCAGTGGTTTTCAGCTGCCAGAGGCCTGAGAGAGTTTGGGCATACTC
8
## 245       DMD CCATTCAGAAGAATGATAAATGCCACAAGCATTTGGAAACAGGCTTCCCT
7
## 322  KIAA0692 AGCTCCACCTTGACCCAGCCTCACAACAAAAAGTTTGTGTATGACCAGGC
9
## 375 LOC202134 AGTGGGCAGAATGATGAGGGAAGTGGGCACGTGCCCATGTTCTTCTTGGC
7
## 382 LOC202134 TTCATCCAGGCCTGCGCCGGTGTTCACAGTGGTCCTCATCTAAGCCAGCC
7
## 591     PLEC1 CCGGGCCTTCTCGTGGTACCCTGCCTGCTGCCTTTGCCCCCGCACTGACT
8
## 47       BDNF GCTCGCTGAAGTTGGCTTCCTAGCGGTGTAGGCTGGAATAGACTCTTGGC
7
## 575     PLEC1 GGCGCAGACATGGACCCCTCGCGAGCCATCCAGAACGAGATCAGCTCCCT
8
```

```
## 313  KIAA0692 GATCACAGGCACAGGGAAGCCACAAGGAGCTCTGTATGAGTTGTGTTTGC
9
## 495 LOC339047 TTTCAGGCCCATGGCAGAGGGTGGGCTCAGGAGGGCCATCGTGGGTGTCC
7
## 170     DDX12 GCAGGGGAGATTGGGTTTAGGGGCTTTCCTGGTCTGCATTCTGCTACAGC
10
## 460 LOC339047 AGTGCCCACATCACACAGCATCTAGCACGTAACTGCACCCCGGGAGTCGT
7
## 437  LOC23117 GGCTCTGTTGGAATCCGCATAGTGTGGAAATGAGTTTGCCCTGGAAAGGG
8
## 502 LOC653086 AGTGTTGGGACTACAGGTGTGTGTTACTGCTCCCAGCTGGGAGGCAGGCT
7
## 509 LOC653086 GTGAGCCTGTTTCATCATCTGTAAACTTTGAATAATGATACCTACCCCGC
7
## 467 LOC339047 CGCCCTGAAAGGACCAGGACATGCGGGTGCGGTGGCTGCTCTTTTGGCTC
7
## 150     DDX12 CAGGGCAGGAACCACGTCTTTACAGTTTGATGTTCCCAGAGCTGACCCAG
10
## 75     CTNNB1 CAGGAATCTAGTCTGGATGACTGCTTCTGGAGCCTGGATGCAGTACCATT
7
## 295  KIAA0692 GTTGTTCTGGACGATCTTCGGGATCCTCTGGGGCACTGTGACACTCGGAG
9
## 180     DDX12 TCTCCTGCCCCCTCCGGAAGCTTGGATGCCCCTCCACACCCTCTTGATCT
10
## 389  LOC23117 CTGGCCTTCCCTCATCAGCCGTAAATGATGATTTACTGCTGTTACCATCA
8
## 103    CTNNB1 GCAATTTGCCAAGTTTCTTTAGCATTTGGCCCTGGATTACGCTGGACCCC
7
## 413  LOC23117 CCCTTCCTACATTCTTGTTTTCATTTTTTCGGAGGAAGAGGAGTTGCTAG
8
## 523 LOC653086 AGCAGCACATCGTCATTTTACAATTGAGAAACATGGAGACTCCAAATGGA
7
## 421  LOC23117 GGGAAGTACATGGGGCAGATGGAAGAACCTGAGATAATCGCAAGGATGGC
8
## 40       BDNF TCAGACCCCTCAGGCCACTGCTGTTCCTGTCACACATTCCTGCAAAGGAC
7
## 607     PLEC1 CTCCGTCTGCCCCGTGGGCTCCTGCCACCGTCCCCGATGAAGATCGTGCC
8
## 453 LOC339047 TTTCCTGAAATGGAGCTTTGCTCTTGTTGCCCAGGCCGTAGTGCAATGGC
7
## 599     PLEC1 GCCTTTGCCTCGCCGAGGGAGGTCTTGCTGGAGCGGCCGTGCTGGCTGGA
8
## 12       BDNF ATGTACGTGGGGGATTCTTGACTCGGGTTAGTCTCTGGGGATGCAGAGCC
7
## 583     PLEC1 CAGCCCTGGGGACACACTGCCCTGGAACCTTGGGAAAACGCAGCGGAGCC
8
## 190     DDX12 CGTTGCTACAAGCTGTTTTTTGAATGTCTCTACACAGTCCAGGCAGGAAG
10
## 481 LOC339047 TCTGTATGGACCCTGCCAAGCTCTGCCCCTCTGCCCCTGCATTGGGGCGC
7
```

```
## 54       BDNF TGGGGAGACGAGATTTTAAGACACTTGAGTCTCCAGGACAGCAAAGGCAC
7
## 160     DDX12 CAGACTTCTCGCTTCCTTTCTGCTGGGCCTCTGAGGGGTCATGGGGCCAT
10
## 304  KIAA0692 AAGTGGTGCCTGGCTGTCCCTATACTGTGCTGCTGGGTGTTCCAGCCTGT
9
## 224       DMD GCAGCCAACTTATTGGCATGATGGAGTGACAGGAAAAACAGCTGGCATGG
7
## 200     DDX12 TTACTGGGGATGGTATTTAGGAGCCAGGAAAGCCGGTGCATTCCTAGTGA
10
## 544 LOC653086 TACCTGGCCTATCTTTCATAGGTTATATAAATTCCTTGGTTCCCAGTTTT
7
## 217       DMD GGGTTTTCTCAGGATTGCTATGCAACAGGATCAGTGCTGTAGTGCCCGGT
7
## 120     DDX12 ACATGTGCTGTCACTGGAACTTGCTCTTTTCACTCAGCAGCCAGAGGGTC
10
## 110     DDX12 CCAGTCCCTGACTACAGAGGATTTCCCCAAAGTCCCTGGCTGTGAGGTTC
10
## 130     DDX12 AAACGTTACAGTGTTCCGATGAGACACAGTAGGCAGTACTTGGGAGGGTC
10
## 368 LOC202134 GACCAAAGCAGGACAATTGCTTGATCCCAGGAGTTTAAGACCAGCCGGGG
7
## 537 LOC653086 AAGGACTCAGATGCAGGGTCTTCTCTGCTCCCCGTCACACAGAGGGTGGC
7
## 277  KIAA0692 CAGGCGACTGGGTAGCAGATGTGGAAGCTGATGGTTAGGCCCAGGGCATG
9
## 488 LOC339047 GACCTGTAGCTAAACCTTCCACCAGCGCTTGAGAACTTAATTTGAACCGG
7
## 238       DMD GCACTCCGACTACATCAGGAGAAGATGTTCGAGACTTTGCCAAGGTACTA
7
## 354 LOC202134 CCACGCCGGCAAAGAAATTGGAAGACTCCACCATTACAGGCAGCCACCAG
7
## 26       BDNF CTTGCTGTGGTCTCTTTGTGGCAGAAGTGTTTCATGCATGGCAGCAGGCC
7
## 567     PLEC1 AGCCTCTGTTCCCCTAGTAAGTGCCTTCCATGTCGGCCTCTAACCCCAGG
8
## 347 LOC202134 CCTGTTTGGATCACATGGTCTTGTCCTGATAACTTGGAAGAGGTTGCTTC
7
## 1       ABCA8 GCCAACCTCCTCTCACAGCCTCTGTATCTCTGCAGGCCATACTGGTTCCA
1
## 3       ADH1A CAGATGTTTTCCCTTGTGGCAGTCTTCAGCCTCCTCTACCCTACATGATC
1
## 6         FOS CCCAGTGACACTTCAGAGAGCTGGTAGTTAGTAGCATGTTGAGCCAGGCC
1
## 7        FOSB TGACTGTCCCTGCCAATGCTCCAGCTGTCGTCTGACTCTGGGTTCGTTGG
1
## 9       KRT19 CAGCTGGGCGATGTGCGAGCTGATAGTGAGCGGCAGAATCAGGAGTACCA
1
##     foldChangeMean_UL_to_nonUL
## 8                   18.0591346
```

```
## 11                  14.9511840
## 2                   11.9638706
## 4                   10.8264232
## 10                  10.7817440
## 68                   1.5689829
## 89                   1.5248323
## 82                   1.2503004
## 231                  1.1977828
## 210                  1.1332249
## 96                   1.1098758
## 140                  1.1076674
## 397                  1.0971923
## 361                  1.0775532
## 286                  1.0762308
## 61                   1.0740152
## 516                  1.0680820
## 33                   1.0451334
## 551                  1.0433765
## 445                  1.0317830
## 474                  1.0291369
## 331                  1.0250419
## 559                  1.0243757
## 530                  1.0237370
## 259                  1.0212595
## 429                  1.0204791
## 252                  1.0202573
## 268                  1.0150804
## 19                   1.0137555
## 340                  1.0132932
## 405                  1.0108892
## 245                  1.0100160
## 322                  1.0098454
## 375                  1.0093062
## 382                  1.0091169
## 591                  1.0071980
## 47                   1.0067778
## 575                  1.0042547
## 313                  1.0038554
## 495                  1.0036545
## 170                  1.0019589
## 460                  1.0005056
## 437                  0.9998782
## 502                  0.9991520
## 509                  0.9982094
## 467                  0.9981707
## 150                  0.9980314
## 75                   0.9977081
## 295                  0.9976800
## 180                  0.9964084
## 389                  0.9937542
```

```
## 103                  0.9930219
## 413                  0.9927896
## 523                  0.9924044
## 421                  0.9921585
## 40                   0.9911345
## 607                  0.9868791
## 453                  0.9867059
## 599                  0.9860355
## 12                   0.9821732
## 583                  0.9816619
## 190                  0.9793547
## 481                  0.9791535
## 54                   0.9767836
## 160                  0.9762911
## 304                  0.9737579
## 224                  0.9735481
## 200                  0.9725752
## 544                  0.9708972
## 217                  0.9699886
## 120                  0.9694418
## 110                  0.9672238
## 130                  0.9664926
## 368                  0.9545894
## 537                  0.9529309
## 277                  0.9406746
## 488                  0.9392117
## 238                  0.9047898
## 354                  0.8995475
## 26                   0.8520654
## 567                  0.7581050
## 347                  0.4746867
## 1                    0.1977871
## 3                    0.1961796
## 6                    0.1860882
## 7                    0.1801972
## 9                    0.1599868
```

```
write.csv(KG1,'keyGenes_UL_FCs_CNVs.csv', row.names=FALSE)
```

Order by gene count, then by fold change.

```
KG2 <- KG1[with(KG1, order(gene_count, foldChangeMean_UL_to_nonUL,
decreasing=TRUE)),]
```

Lets add in a fold change of the median value ratios of UL to non-UL samples to compare.

```
colnames(KG2)[18] <- 'foldChange_Mean'
KG2$foldChange_Median <- KG2$UL_Median/KG2$nonUL_Median
```

Lets look at some of these copy number variants of one gene with seven copy number variants or CNVs and see where the changes in the nucleotide sequences occur. Copy

number variations in nucleotides can have short repeats, jumps in sequence, insertions, or deletions of a gene. I have been calling these CNVs genotypes, which are the traits and alleles responsible for the physical traits or phenotypes of an organism. Some CNVs are responsible for diseases, and in tumors there are many different CNVs that are found to be responsible. A uterine leiomyoma or fibroid is a benign tumor. These samples were taken from uterus tissue with these uternine tumors and the same neighboring uterine tissue without uterine tumors.

```
CTNNB1 <- subset(KG2, KG2$Symbol=='CTNNB1')
CTNNB1_seq <- CTNNB1[,1:2]
```

Add in a column to describe the length of the nucleotides in each copy number variant nucleotide strand.

```
CTNNB1_seq$SEQUENCE <- as.character(CTNNB1$SEQUENCE)

CTNNB1_seq$nChar <- nchar(CTNNB1_seq$SEQUENCE)
```

Lets look at the CNVs of the CTNNB1 gene.

```
CTNNB1_seq
```

```
##     Symbol                                                SEQUENCE nChar
## 68  CTNNB1 AGCTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAG    50
## 89  CTNNB1 CTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAGCT    50
## 82  CTNNB1 AGTCTCTCGTAGTGTTAAGTTATAGTGAATACTGCTACAGCAATTTCTAA    50
## 96  CTNNB1 GCCTCTTGCACTCTGAATTGGGAATGTTTGCACCACAGTGGGGGGCTTGC    50
## 61  CTNNB1 CAAACTTTACAGAGGAGAATGCCCTGTTTGTTAACCATGTTTCTTTTGGC    50
## 75  CTNNB1 CAGGAATCTAGTCTGGATGACTGCTTCTGGAGCCTGGATGCAGTACCATT    50
## 103 CTNNB1 GCAATTTGCCAAGTTTCTTTAGCATTTGGCCCTGGATTACGCTGGACCCC    50
```

From the above, some of the CNVs make you wonder if they are even the same gene. The first two have the same pattern of 'CTGCAGGG' then some variations. Then its not obvious what the other sequence alignments are. We could go back to the cytoband location and where the gene starts to see if there is more information.

Lets get the SEQUENCE, protein product, and cytoband columns from the original UL1 table.

```
cytoband <- UL1[,c(15,20,24)]
```

Now combine with the CTNNB1_seq and the KG2 table.

```
CTNNB1_cyto <- merge(cytoband, CTNNB1_seq, by.x='SEQUENCE', by.y='SEQUENCE')
KG2_cyto <- merge(cytoband, KG2, by.x='SEQUENCE', by.y='SEQUENCE')
```

Now lets look at the KG2_cyto table to see where these CNVs are located within the cytoband of each gene location.

```
KG3 <- KG2_cyto[with(KG2_cyto, order(gene_count,foldChange_Mean, decreasing =
TRUE)),]
KG3[,1:5]
```

```
##                                                 SEQUENCE Protein_Product
## 37 CCGCCGGGCTGCTTTTTCCTTGGATGCCCATCAGGACGCCTCAGTTCTCT     XP_936926.1
## 56 GCAGGGGAGATTGGGTTTAGGGGCTTTCCTGGTCTGCATTCTGCTACAGC     XP_937020.1
## 26 CAGGGCAGGAACCACGTCTTTACAGTTTGATGTTCCCAGAGCTGACCCAG     XP_936919.1
## 77 TCTCCTGCCCCCTCCGGAAGCTTGGATGCCCCTCCACACCCTCTTGATCT     XP_936947.1
## 41 CGTTGCTACAAGCTGTTTTTTGAATGTCTCTACACAGTCCAGGCAGGAAG     XP_937000.1
## 20 CAGACTTCTCGCTTCCTTTCTGCTGGGCCTCTGAGGGGTCATGGGGCCAT     XP_936988.1
## 84 TTACTGGGGATGGTATTTAGGAGCCAGGAAAGCCGGTGCATTCCTAGTGA     XP_936932.1
## 6  ACATGTGCTGTCACTGGAACTTGCTCTTTTCACTCAGCAGCCAGAGGGTC     XP_936976.1
## 30 CCAGTCCCTGACTACAGAGGATTTCCCCAAAGTCCCTGGCTGTGAGGTTC     XP_936952.1
## 1  AAACGTTACAGTGTTCCGATGAGACACAGTAGGCAGTACTTGGGAGGGTC     XP_936980.1
## 72 TAAGTGCAGTGAGCTCTGGCGGAAACCACCCTCTGCCCCGTCTGTTGGAT     XP_935983.1
## 28 CATTGTAATGATAAGGAAATGTTGCGATCAAATAAGATTTAGACACACTT     XP_935991.1
## 49 GAGTGCTGGGAAGGTTAATGTTAAATGGGTTGTGTGTCGGGGAGGGTACA     XP_935974.1
## 51 GCAAATGTAACTCAGGGGTTTGGGGCCAGAGGAAGAGGGAGAAGGTGGCC     XP_935936.1
## 10 AGCTCCACCTTGACCCAGCCTCACAACAAAAAGTTTGTGTATGACCAGGC     XP_935967.1
## 50 GATCACAGGCACAGGGAAGCCACAAGGAGCTCTGTATGAGTTGTGTTTGC     XP_935893.1
## 71 GTTGTTCTGGACGATCTTCGGGATCCTCTGGGGCACTGTGACACTCGGAG     XP_936004.1
## 4  AAGTGGTGCCTGGCTGTCCCTATACTGTGCTGCTGGGTGTTCCAGCCTGT     XP_935903.1
## 25 CAGGCGACTGGGTAGCAGATGTGGAAGCTGATGGTTAGGCCCAGGGCATG     XP_935881.1
## 74 TCAACCACATCCTTCAAAAGGACTATGCCTGTTTATAAGCCCAGCTGTTT     XP_938957.1
## 33 CCCGACGAGCAGGACTTCATCCAGGCCTACGAGGAGGTGCGCGAGAAGTA     NP_958780.1
## 83 TGTCGTTTCCTCCATTCTTCACCAAAACATCAGCGTACATAGGCACATGG     XP_938806.1
## 35 CCCTCGGGCAGCCTGTTTCCCTCCCTGGTGGTTGTGGGTCACGTTGTCAC     NP_958784.1
## 64 GGCTCCTCTTTGGGCTCCTACTGGAATTTATCAGCCATCAGTGCATCTCT     XP_938917.1
## 2  AAAGCAGTGGTTTTCAGCTGCCAGAGGCCTGAGAGAGTTTGGGCATACTC     XP_938927.1
## 38 CCGGGCCTTCTCGTGGTACCCTGCCTGCTGCCTTTGCCCCCGCACTGACT     NP_958782.1
## 63 GGCGCAGACATGGACCCCTCGCGAGCCATCCAGAACGAGATCAGCTCCCT     NP_958781.1
## 65 GGCTCTGTTGGAATCCGCATAGTGTGGAAATGAGTTTGCCCTGGAAAGGG     XP_938916.1
## 45 CTGGCCTTCCCTCATCAGCCGTAAATGATGATTTACTGCTGTTACCATCA     XP_939002.1
## 36 CCCTTCCTACATTCTTGTTTTCATTTTTTCGGAGGAAGAGGAGTTGCTAG     XP_938960.1
## 66 GGGAAGTACATGGGGCAGATGGAAGAACCTGAGATAATCGCAAGGATGGC     XP_938807.1
## 42 CTCCGTCTGCCCCGTGGGCTCCTGCCACCGTCCCCGATGAAGATCGTGCC     NP_958783.1
## 61 GCCTTTGCCTCGCCGAGGGAGGTCTTGCTGGAGCGGCCGTGCTGGCTGGA     NP_958785.1
## 22 CAGCCCTGGGGACACACTGCCCTGGAACCTTGGGAAAACGCAGCGGAGCC     NP_000436.2
## 9  AGCCTCTGTTCCCCTAGTAAGTGCCTTCCATGTCGGCCTCTAACCCCAGG     NP_958786.1
## 11 AGCTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAG     XP_950743.1
## 44 CTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAGCT     NP_001895.1
## 13 AGTCTCTCGTAGTGTTAAGTTATAGTGAATACTGCTACAGCAATTTCTAA     NP_001895.1
## 27 CAGTGTTGGGATCACTCACTTTCCCCCTACAGGACTCAGATCTGGGAGGC     NP_003997.1
## 43 CTCCTCTCAGCTGAACACCCTCCTTTCACTCCCAAATGCAAACAGTCTCT     NP_004010.1
## 60 GCCTCTTGCACTCTGAATTGGGAATGTTTGCACCACAGTGGGGGGCTTGC     XP_950747.1
## 57 GCCAAAGGAATGGGCTCCAGACACCCCCTCTTCCAGAGCAAGGATGAAGG     XP_937236.1
## 19 CAAACTTTACAGAGGAGAATGCCCTGTTTGTTAACCATGTTTCTTTTGGC     XP_950746.1
## 80 TGATGTGTCACGCCACTGTACTCCAGCCTGACGGCAGAGCGAGACTCCAT     XP_936088.1
## 34 CCCTCCACCTCCTGCTCGGGGGGCTTTAATGAGACACCCACCGCTGCTGT     NP_001700.2
```

```
## 7  ACTGCCTGTGTGGCTCCTTGAGTGCGCGGAGGCCAAAGCTGAGATGACTT     XP_937640.1
## 69 GGTGTGCTCTGGTATGTAATGACAATATGTGAACAAACCTGTGGAATTAA     XP_936056.1
## 76 TCTATCAACAGAGCTGAATGAGTGCCAGGAAGCTGCGAAATCTGTCTTAC     NP_004003.1
## 5  AATAATAGAGTGTGGGAGTTTTGGGGCCGAAGTCTTTCCCGGAGCAGCTG     NP_733929.1
## 18 CAAACCCTTGAAGACATTTCAGGGCCATGCTCACTTGGGAGGGTTTGAGG     XP_937228.1
## 31 CCATTCAGAAGAATGATAAATGCCACAAGCATTTGGAAACAGGCTTCCCT     NP_004001.1
## 15 AGTGGGCAGAATGATGAGGGAAGTGGGCACGTGCCCATGTTCTTCTTGGC     XP_937222.1
## 85 TTCATCCAGGCCTGCGCCGGTGTTCACAGTGGTCCTCATCTAAGCCAGCC     XP_937214.1
## 62 GCTCGCTGAAGTTGGCTTCCTAGCGGTGTAGGCTGGAATAGACTCTTGGC     NP_733928.1
## 86 TTTCAGGCCCATGGCAGAGGGTGGGCTCAGGAGGGCCATCGTGGGTGTCC     XP_937694.1
## 14 AGTGCCCACATCACACAGCATCTAGCACGTAACTGCACCCCGGGAGTCGT     XP_937456.1
## 16 AGTGTTGGGACTACAGGTGTGTGTTACTGCTCCCAGCTGGGAGGCAGGCT     XP_936104.1
## 70 GTGAGCCTGTTTCATCATCTGTAAACTTTGAATAATGATACCTACCCCGC     XP_936038.1
## 40 CGCCCTGAAAGGACCAGGACATGCGGGTGCGGTGGCTGCTCTTTTGGCTC     XP_937724.1
## 24 CAGGAATCTAGTCTGGATGACTGCTTCTGGAGCCTGGATGCAGTACCATT     XP_950748.1
## 53 GCAATTTGCCAAGTTTCTTTAGCATTTGGCCCTGGATTACGCTGGACCCC     XP_947138.1
## 8  AGCAGCACATCGTCATTTTACAATTGAGAAACATGGAGACTCCAAATGGA     XP_936080.1
## 75 TCAGACCCCTCAGGCCACTGCTGTTCCTGTCACACATTCCTGCAAAGGAC     NP_733931.1
## 87 TTTCCTGAAATGGAGCTTTGCTCTTGTTGCCCAGGCCGTAGTGCAATGGC     XP_937537.1
## 17 ATGTACGTGGGGGATTCTTGACTCGGGTTAGTCTCTGGGGATGCAGAGCC     NP_733930.1
## 78 TCTGTATGGACCCTGCCAAGCTCTGCCCCTCTGCCCCTGCATTGGGGCGC     XP_937505.1
## 82 TGGGGAGACGAGATTTTAAGACACTTGAGTCTCCAGGACAGCAAAGGCAC     NP_733927.1
## 55 GCAGCCAACTTATTGGCATGATGGAGTGACAGGAAAAACAGCTGGCATGG     NP_000100.2
## 73 TACCTGGCCTATCTTTCATAGGTTATATAAATTCCTTGGTTCCCAGTTTT     XP_936049.1
## 67 GGGTTTTCTCAGGATTGCTATGCAACAGGATCAGTGCTGTAGTGCCCGGT     NP_004005.1
## 47 GACCAAAGCAGGACAATTGCTTGATCCCAGGAGTTTAAGACCAGCCGGGG     XP_932593.1
## 3  AAGGACTCAGATGCAGGGTCTTCTCTGCTCCCCGTCACACAGAGGGTGGC     XP_936046.1
## 48 GACCTGTAGCTAAACCTTCCACCAGCGCTTGAGAACTTAATTTGAACCGG     XP_937490.1
## 54 GCACTCCGACTACATCAGGAGAAGATGTTCGAGACTTTGCCAAGGTACTA     NP_004010.1
## 29 CCACGCCGGCAAAGAAATTGGAAGACTCCACCATTACAGGCAGCCACCAG     XP_937214.1
## 46 CTTGCTGTGGTCTCTTTGTGGCAGAAGTGTTTCATGCATGGCAGCAGGCC     NP_001700.2
## 39 CCTGTTTGGATCACATGGTCTTGTCCTGATAACTTGGAAGAGGTTGCTTC     XP_371783.3
## 81 TGCGAGACCTGGGTGTCCAACCTGCGCTACAACCACATGCTGCGGAAGAA     NP_003827.3
## 52 GCAACGCTCCTCTGAAATGCTTGTCTTTTTTCTGTTGCCGAAATAGCTGG     NP_061159.1
## 59 GCCCCAGCAAGCCTCCCTCCATCCTCCAGTGGGAAACTGTTGATGGTGTT     NP_006202.1
## 68 GGTATTGCTGATCGTATGCAGAAGGAAATCACTGCTCTGGCTCCTAGCAC     NP_005150.1
## 12 AGGCCCTGGAGGCTGCAACATACCTCAATCCTGTCCCAGGCCGGATCCTC     NP_005931.2
## 58 GCCAACCTCCTCTCACAGCCTCTGTATCTCTGCAGGCCATACTGGTTCCA     NP_009099.1
## 21 CAGATGTTTTCCCTTGTGGCAGTCTTCAGCCTCCTCTACCCTACATGATC     NP_000658.1
## 32 CCCAGTGACACTTCAGAGAGCTGGTAGTTAGTAGCATGTTGAGCCAGGCC     NP_005243.1
## 79 TGACTGTCCCTGCCAATGCTCCAGCTGTCGTCTGACTCTGGGTTCGTTGG     NP_006723.1
## 23 CAGCTGGGCGATGTGCGAGCTGATAGTGAGCGGCAGAATCAGGAGTACCA     NP_002267.2
##          Cytoband    Symbol gene_count
## 37      12p13.31a     DDX12         10
## 56      12p13.31a     DDX12         10
## 26      12p13.31a     DDX12         10
## 77      12p13.31a     DDX12         10
## 41      12p13.31a     DDX12         10
## 20      12p13.31a     DDX12         10
## 84      12p13.31a     DDX12         10
```

```
## 6       12p13.31a     DDX12         10
## 30      12p13.31a     DDX12         10
## 1       12p13.31a     DDX12         10
## 72      12q24.33d  KIAA0692          9
## 28      12q24.33d  KIAA0692          9
## 49      12q24.33d  KIAA0692          9
## 51      12q24.33d  KIAA0692          9
## 10      12q24.33d  KIAA0692          9
## 50      12q24.33d  KIAA0692          9
## 71      12q24.33d  KIAA0692          9
## 4       12q24.33d  KIAA0692          9
## 25      12q24.33d  KIAA0692          9
## 74       16p12.2a  LOC23117          8
## 33        8q24.3g     PLEC1          8
## 83       16p12.2a  LOC23117          8
## 35        8q24.3g     PLEC1          8
## 64       16p12.2a  LOC23117          8
## 2        16p12.2a  LOC23117          8
## 38        8q24.3g     PLEC1          8
## 63        8q24.3g     PLEC1          8
## 65       16p12.2a  LOC23117          8
## 45       16p12.2a  LOC23117          8
## 36       16p12.2a  LOC23117          8
## 66       16p12.2a  LOC23117          8
## 42        8q24.3g     PLEC1          8
## 61        8q24.3g     PLEC1          8
## 22        8q24.3g     PLEC1          8
## 9         8q24.3g     PLEC1          8
## 11        3p22.1b    CTNNB1          7
## 44        3p22.1b    CTNNB1          7
## 13        3p22.1b    CTNNB1          7
## 27 Xp21.2a-p21.1d       DMD          7
## 43 Xp21.2a-p21.1d       DMD          7
## 60        3p22.1b    CTNNB1          7
## 57        5q35.2d LOC202134          7
## 19        3p22.1b    CTNNB1          7
## 80             NA LOC653086          7
## 34       11p14.1d      BDNF          7
## 7       16p13.11b LOC339047          7
## 69             NA LOC653086          7
## 76 Xp21.2a-p21.1d       DMD          7
## 5        11p14.1d      BDNF          7
## 18        5q35.2d LOC202134          7
## 31 Xp21.2a-p21.1d       DMD          7
## 15        5q35.2d LOC202134          7
## 85        5q35.2d LOC202134          7
## 62       11p14.1d      BDNF          7
## 86      16p13.11b LOC339047          7
## 14      16p13.11b LOC339047          7
## 16             NA LOC653086          7
```

```
## 70             NA LOC653086          7
## 40      16p13.11b LOC339047          7
## 24        3p22.1b    CTNNB1          7
## 53        3p22.1b    CTNNB1          7
## 8              NA LOC653086          7
## 75       11p14.1d      BDNF          7
## 87      16p13.11b LOC339047          7
## 17       11p14.1d      BDNF          7
## 78      16p13.11b LOC339047          7
## 82       11p14.1d      BDNF          7
## 55 Xp21.2a-p21.1d       DMD          7
## 73             NA LOC653086          7
## 67 Xp21.2a-p21.1d       DMD          7
## 47        5q35.2d LOC202134          7
## 3              NA LOC653086          7
## 48      16p13.11b LOC339047          7
## 54 Xp21.2a-p21.1d       DMD          7
## 29        5q35.2d LOC202134          7
## 46       11p14.1d      BDNF          7
## 39        5q35.2d LOC202134          7
## 81       14q32.2b      DLK1          2
## 52       15q25.1b  KIAA1199          1
## 59        8q12.1b      PENK          1
## 68         15q14a      ACTC          1
## 12      22q11.23a     MMP11          1
## 58       17q24.2c     ABCA8          1
## 21          4q23b     ADH1A          1
## 32       14q24.3b       FOS          1
## 79      19q13.32a      FOSB          1
## 23       17q21.2b     KRT19          1
```

```
CTNNB1_b <- subset(KG3, KG3$Symbol=='CTNNB1')
CTNNB1_b[,c(1:5,20:21)]
```

```
##                                                SEQUENCE Protein_Product
Cytoband
## 11 AGCTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAG     XP_950743.1
3p22.1b
## 44 CTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAGCT     NP_001895.1
3p22.1b
## 13 AGTCTCTCGTAGTGTTAAGTTATAGTGAATACTGCTACAGCAATTTCTAA     NP_001895.1
3p22.1b
## 60 GCCTCTTGCACTCTGAATTGGGAATGTTTGCACCACAGTGGGGGGCTTGC     XP_950747.1
3p22.1b
## 19 CAAACTTTACAGAGGAGAATGCCCTGTTTGTTAACCATGTTTCTTTTGGC     XP_950746.1
3p22.1b
## 24 CAGGAATCTAGTCTGGATGACTGCTTCTGGAGCCTGGATGCAGTACCATT     XP_950748.1
3p22.1b
## 53 GCAATTTGCCAAGTTTCTTTAGCATTTGGCCCTGGATTACGCTGGACCCC     XP_947138.1
3p22.1b
```

```
##    Symbol gene_count foldChange_Mean foldChange_Median
## 11 CTNNB1          7       1.5689829         1.5744314
## 44 CTNNB1          7       1.5248323         1.4655024
## 13 CTNNB1          7       1.2503004         1.1602376
## 60 CTNNB1          7       1.1098758         1.0850642
## 19 CTNNB1          7       1.0740152         1.0471422
## 24 CTNNB1          7       0.9977081         0.9924948
## 53 CTNNB1          7       0.9930219         0.9818097
```

The cytoband location of each of these CNVs for CTNNB1 is the same location on chromosome 3 on the p strand/direction along 22.1b. Also, the fold change for the mean and median values for the first listed CNVs changed by 16-57 percent more in UL compared to non-UL samples. This could mean that these four CNVs of the gene CTNNB1 offer some clues as to what mutations or changes impact risk in developing uterine leiomyomas for some females.

Lets order the key genes by fold change median then by CNVs.

```
KG4 <- KG3[with(KG3, order(foldChange_Median, gene_count, decreasing =
TRUE)),]
KG4[,c(1:5,21)]
```

```
##                                                SEQUENCE Protein_Product
## 52 GCAACGCTCCTCTGAAATGCTTGTCTTTTTTCTGTTGCCGAAATAGCTGG     NP_061159.1
## 68 GGTATTGCTGATCGTATGCAGAAGGAAATCACTGCTCTGGCTCCTAGCAC     NP_005150.1
## 12 AGGCCCTGGAGGCTGCAACATACCTCAATCCTGTCCCAGGCCGGATCCTC     NP_005931.2
## 81 TGCGAGACCTGGGTGTCCAACCTGCGCTACAACCACATGCTGCGGAAGAA     NP_003827.3
## 59 GCCCCAGCAAGCCTCCCTCCATCCTCCAGTGGGAAACTGTTGATGGTGTT     NP_006202.1
## 11 AGCTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAG     XP_950743.1
## 44 CTGCAGGGGTCCTCTGTGAACTTGCTCAGGACAAGGAAGCTGCAGAAGCT     NP_001895.1
## 43 CTCCTCTCAGCTGAACACCCTCCTTTCACTCCCAAATGCAAACAGTCTCT     NP_004010.1
## 13 AGTCTCTCGTAGTGTTAAGTTATAGTGAATACTGCTACAGCAATTTCTAA     NP_001895.1
## 27 CAGTGTTGGGATCACTCACTTTCCCCCTACAGGACTCAGATCTGGGAGGC     NP_003997.1
## 60 GCCTCTTGCACTCTGAATTGGGAATGTTTGCACCACAGTGGGGGGCTTGC     XP_950747.1
## 74 TCAACCACATCCTTCAAAAGGACTATGCCTGTTTATAAGCCCAGCTGTTT     XP_938957.1
## 10 AGCTCCACCTTGACCCAGCCTCACAACAAAAAGTTTGTGTATGACCAGGC     XP_935967.1
## 19 CAAACTTTACAGAGGAGAATGCCCTGTTTGTTAACCATGTTTCTTTTGGC     XP_950746.1
## 33 CCCGACGAGCAGGACTTCATCCAGGCCTACGAGGAGGTGCGCGAGAAGTA     NP_958780.1
## 80 TGATGTGTCACGCCACTGTACTCCAGCCTGACGGCAGAGCGAGACTCCAT     XP_936088.1
## 31 CCATTCAGAAGAATGATAAATGCCACAAGCATTTGGAAACAGGCTTCCCT     NP_004001.1
## 64 GGCTCCTCTTTGGGCTCCTACTGGAATTTATCAGCCATCAGTGCATCTCT     XP_938917.1
## 69 GGTGTGCTCTGGTATGTAATGACAATATGTGAACAAACCTGTGGAATTAA     XP_936056.1
## 28 CATTGTAATGATAAGGAAATGTTGCGATCAAATAAGATTTAGACACACTT     XP_935991.1
## 76 TCTATCAACAGAGCTGAATGAGTGCCAGGAAGCTGCGAAATCTGTCTTAC     NP_004003.1
## 16 AGTGTTGGGACTACAGGTGTGTGTTACTGCTCCCAGCTGGGAGGCAGGCT     XP_936104.1
## 37 CCGCCGGGCTGCTTTTTCCTTGGATGCCCATCAGGACGCCTCAGTTCTCT     XP_936926.1
## 86 TTTCAGGCCCATGGCAGAGGGTGGGCTCAGGAGGGCCATCGTGGGTGTCC     XP_937694.1
## 49 GAGTGCTGGGAAGGTTAATGTTAAATGGGTTGTGTGTCGGGGAGGGTACA     XP_935974.1
## 83 TGTCGTTTCCTCCATTCTTCACCAAAACATCAGCGTACATAGGCACATGG     XP_938806.1
## 35 CCCTCGGGCAGCCTGTTTCCCTCCCTGGTGGTTGTGGGTCACGTTGTCAC     NP_958784.1
```

```
## 57 GCCAAAGGAATGGGCTCCAGACACCCCCTCTTCCAGAGCAAGGATGAAGG     XP_937236.1
## 14 AGTGCCCACATCACACAGCATCTAGCACGTAACTGCACCCCGGGAGTCGT     XP_937456.1
## 50 GATCACAGGCACAGGGAAGCCACAAGGAGCTCTGTATGAGTTGTGTTTGC     XP_935893.1
## 70 GTGAGCCTGTTTCATCATCTGTAAACTTTGAATAATGATACCTACCCCGC     XP_936038.1
## 38 CCGGGCCTTCTCGTGGTACCCTGCCTGCTGCCTTTGCCCCCGCACTGACT     NP_958782.1
## 65 GGCTCTGTTGGAATCCGCATAGTGTGGAAATGAGTTTGCCCTGGAAAGGG     XP_938916.1
## 78 TCTGTATGGACCCTGCCAAGCTCTGCCCCTCTGCCCCTGCATTGGGGCGC     XP_937505.1
## 85 TTCATCCAGGCCTGCGCCGGTGTTCACAGTGGTCCTCATCTAAGCCAGCC     XP_937214.1
## 7  ACTGCCTGTGTGGCTCCTTGAGTGCGCGGAGGCCAAAGCTGAGATGACTT     XP_937640.1
## 5  AATAATAGAGTGTGGGAGTTTTGGGGCCGAAGTCTTTCCCGGAGCAGCTG     NP_733929.1
## 18 CAAACCCTTGAAGACATTTCAGGGCCATGCTCACTTGGGAGGGTTTGAGG     XP_937228.1
## 62 GCTCGCTGAAGTTGGCTTCCTAGCGGTGTAGGCTGGAATAGACTCTTGGC     NP_733928.1
## 66 GGGAAGTACATGGGGCAGATGGAAGAACCTGAGATAATCGCAAGGATGGC     XP_938807.1
## 41 CGTTGCTACAAGCTGTTTTTTGAATGTCTCTACACAGTCCAGGCAGGAAG     XP_937000.1
## 45 CTGGCCTTCCCTCATCAGCCGTAAATGATGATTTACTGCTGTTACCATCA     XP_939002.1
## 51 GCAAATGTAACTCAGGGGTTTGGGGCCAGAGGAAGAGGGAGAAGGTGGCC     XP_935936.1
## 40 CGCCCTGAAAGGACCAGGACATGCGGGTGCGGTGGCTGCTCTTTTGGCTC     XP_937724.1
## 84 TTACTGGGGATGGTATTTAGGAGCCAGGAAAGCCGGTGCATTCCTAGTGA     XP_936932.1
## 17 ATGTACGTGGGGGATTCTTGACTCGGGTTAGTCTCTGGGGATGCAGAGCC     NP_733930.1
## 20 CAGACTTCTCGCTTCCTTTCTGCTGGGCCTCTGAGGGGTCATGGGGCCAT     XP_936988.1
## 24 CAGGAATCTAGTCTGGATGACTGCTTCTGGAGCCTGGATGCAGTACCATT     XP_950748.1
## 36 CCCTTCCTACATTCTTGTTTTCATTTTTTCGGAGGAAGAGGAGTTGCTAG     XP_938960.1
## 15 AGTGGGCAGAATGATGAGGGAAGTGGGCACGTGCCCATGTTCTTCTTGGC     XP_937222.1
## 26 CAGGGCAGGAACCACGTCTTTACAGTTTGATGTTCCCAGAGCTGACCCAG     XP_936919.1
## 2  AAAGCAGTGGTTTTCAGCTGCCAGAGGCCTGAGAGAGTTTGGGCATACTC     XP_938927.1
## 75 TCAGACCCCTCAGGCCACTGCTGTTCCTGTCACACATTCCTGCAAAGGAC     NP_733931.1
## 56 GCAGGGGAGATTGGGTTTAGGGGCTTTCCTGGTCTGCATTCTGCTACAGC     XP_937020.1
## 55 GCAGCCAACTTATTGGCATGATGGAGTGACAGGAAAAACAGCTGGCATGG     NP_000100.2
## 8  AGCAGCACATCGTCATTTTACAATTGAGAAACATGGAGACTCCAAATGGA     XP_936080.1
## 42 CTCCGTCTGCCCCGTGGGCTCCTGCCACCGTCCCCGATGAAGATCGTGCC     NP_958783.1
## 53 GCAATTTGCCAAGTTTCTTTAGCATTTGGCCCTGGATTACGCTGGACCCC     XP_947138.1
## 61 GCCTTTGCCTCGCCGAGGGAGGTCTTGCTGGAGCGGCCGTGCTGGCTGGA     NP_958785.1
## 72 TAAGTGCAGTGAGCTCTGGCGGAAACCACCCTCTGCCCCGTCTGTTGGAT     XP_935983.1
## 71 GTTGTTCTGGACGATCTTCGGGATCCTCTGGGGCACTGTGACACTCGGAG     XP_936004.1
## 22 CAGCCCTGGGGACACACTGCCCTGGAACCTTGGGAAAACGCAGCGGAGCC     NP_000436.2
## 73 TACCTGGCCTATCTTTCATAGGTTATATAAATTCCTTGGTTCCCAGTTTT     XP_936049.1
## 77 TCTCCTGCCCCCTCCGGAAGCTTGGATGCCCCTCCACACCCTCTTGATCT     XP_936947.1
## 63 GGCGCAGACATGGACCCCTCGCGAGCCATCCAGAACGAGATCAGCTCCCT     NP_958781.1
## 67 GGGTTTTCTCAGGATTGCTATGCAACAGGATCAGTGCTGTAGTGCCCGGT     NP_004005.1
## 82 TGGGGAGACGAGATTTTAAGACACTTGAGTCTCCAGGACAGCAAAGGCAC     NP_733927.1
## 4  AAGTGGTGCCTGGCTGTCCCTATACTGTGCTGCTGGGTGTTCCAGCCTGT     XP_935903.1
## 48 GACCTGTAGCTAAACCTTCCACCAGCGCTTGAGAACTTAATTTGAACCGG     XP_937490.1
## 87 TTTCCTGAAATGGAGCTTTGCTCTTGTTGCCCAGGCCGTAGTGCAATGGC     XP_937537.1
## 6  ACATGTGCTGTCACTGGAACTTGCTCTTTTCACTCAGCAGCCAGAGGGTC     XP_936976.1
## 34 CCCTCCACCTCCTGCTCGGGGGGCTTTAATGAGACACCCACCGCTGCTGT     NP_001700.2
## 30 CCAGTCCCTGACTACAGAGGATTTCCCCAAAGTCCCTGGCTGTGAGGTTC     XP_936952.1
## 1  AAACGTTACAGTGTTCCGATGAGACACAGTAGGCAGTACTTGGGAGGGTC     XP_936980.1
## 3  AAGGACTCAGATGCAGGGTCTTCTCTGCTCCCCGTCACACAGAGGGTGGC     XP_936046.1
## 47 GACCAAAGCAGGACAATTGCTTGATCCCAGGAGTTTAAGACCAGCCGGGG     XP_932593.1
## 46 CTTGCTGTGGTCTCTTTGTGGCAGAAGTGTTTCATGCATGGCAGCAGGCC     NP_001700.2
```

```
## 29 CCACGCCGGCAAAGAAATTGGAAGACTCCACCATTACAGGCAGCCACCAG     XP_937214.1
## 25 CAGGCGACTGGGTAGCAGATGTGGAAGCTGATGGTTAGGCCCAGGGCATG     XP_935881.1
## 54 GCACTCCGACTACATCAGGAGAAGATGTTCGAGACTTTGCCAAGGTACTA     NP_004010.1
## 9  AGCCTCTGTTCCCCTAGTAAGTGCCTTCCATGTCGGCCTCTAACCCCAGG     NP_958786.1
## 39 CCTGTTTGGATCACATGGTCTTGTCCTGATAACTTGGAAGAGGTTGCTTC     XP_371783.3
## 21 CAGATGTTTTCCCTTGTGGCAGTCTTCAGCCTCCTCTACCCTACATGATC     NP_000658.1
## 58 GCCAACCTCCTCTCACAGCCTCTGTATCTCTGCAGGCCATACTGGTTCCA     NP_009099.1
## 32 CCCAGTGACACTTCAGAGAGCTGGTAGTTAGTAGCATGTTGAGCCAGGCC     NP_005243.1
## 79 TGACTGTCCCTGCCAATGCTCCAGCTGTCGTCTGACTCTGGGTTCGTTGG     NP_006723.1
## 23 CAGCTGGGCGATGTGCGAGCTGATAGTGAGCGGCAGAATCAGGAGTACCA     NP_002267.2
##          Cytoband    Symbol gene_count foldChange_Median
## 52       15q25.1b  KIAA1199          1        21.9688396
## 68         15q14a      ACTC          1        14.6883331
## 12      22q11.23a     MMP11          1        11.8480229
## 81       14q32.2b      DLK1          2         4.7325319
## 59        8q12.1b      PENK          1         3.3078453
## 11        3p22.1b    CTNNB1          7         1.5744314
## 44        3p22.1b    CTNNB1          7         1.4655024
## 43 Xp21.2a-p21.1d       DMD          7         1.2286298
## 13        3p22.1b    CTNNB1          7         1.1602376
## 27 Xp21.2a-p21.1d       DMD          7         1.1029701
## 60        3p22.1b    CTNNB1          7         1.0850642
## 74       16p12.2a  LOC23117          8         1.0667330
## 10      12q24.33d  KIAA0692          9         1.0473309
## 19        3p22.1b    CTNNB1          7         1.0471422
## 33        8q24.3g     PLEC1          8         1.0462839
## 80             NA LOC653086          7         1.0414456
## 31 Xp21.2a-p21.1d       DMD          7         1.0377049
## 64       16p12.2a  LOC23117          8         1.0286458
## 69             NA LOC653086          7         1.0279570
## 28      12q24.33d  KIAA0692          9         1.0247769
## 76 Xp21.2a-p21.1d       DMD          7         1.0233573
## 16             NA LOC653086          7         1.0191080
## 37      12p13.31a     DDX12         10         1.0179040
## 86      16p13.11b LOC339047          7         1.0175470
## 49      12q24.33d  KIAA0692          9         1.0151057
## 83       16p12.2a  LOC23117          8         1.0146392
## 35        8q24.3g     PLEC1          8         1.0138161
## 57        5q35.2d LOC202134          7         1.0137122
## 14      16p13.11b LOC339047          7         1.0120598
## 50      12q24.33d  KIAA0692          9         1.0118272
## 70             NA LOC653086          7         1.0087172
## 38        8q24.3g     PLEC1          8         1.0081202
## 65       16p12.2a  LOC23117          8         1.0071909
## 78      16p13.11b LOC339047          7         1.0071241
## 85        5q35.2d LOC202134          7         1.0066428
## 7       16p13.11b LOC339047          7         1.0047835
## 5        11p14.1d      BDNF          7         1.0046246
## 18        5q35.2d LOC202134          7         1.0037413
## 62       11p14.1d      BDNF          7         1.0016629
```

```
## 66       16p12.2a  LOC23117          8         1.0009319
## 41      12p13.31a     DDX12         10         0.9972581
## 45       16p12.2a  LOC23117          8         0.9970746
## 51      12q24.33d  KIAA0692          9         0.9962676
## 40      16p13.11b LOC339047          7         0.9950549
## 84      12p13.31a     DDX12         10         0.9945794
## 17       11p14.1d      BDNF          7         0.9937282
## 20      12p13.31a     DDX12         10         0.9930535
## 24        3p22.1b    CTNNB1          7         0.9924948
## 36       16p12.2a  LOC23117          8         0.9922770
## 15        5q35.2d LOC202134          7         0.9917228
## 26      12p13.31a     DDX12         10         0.9915254
## 2        16p12.2a  LOC23117          8         0.9908288
## 75       11p14.1d      BDNF          7         0.9900315
## 56      12p13.31a     DDX12         10         0.9899665
## 55 Xp21.2a-p21.1d       DMD          7         0.9894561
## 8               NA LOC653086          7         0.9847075
## 42        8q24.3g     PLEC1          8         0.9844314
## 53        3p22.1b    CTNNB1          7         0.9818097
## 61        8q24.3g     PLEC1          8         0.9800936
## 72      12q24.33d  KIAA0692          9         0.9784906
## 71      12q24.33d  KIAA0692          9         0.9774402
## 22        8q24.3g     PLEC1          8         0.9773820
## 73              NA LOC653086          7         0.9770270
## 77      12p13.31a     DDX12         10         0.9757264
## 63        8q24.3g     PLEC1          8         0.9754829
## 67 Xp21.2a-p21.1d       DMD          7         0.9733874
## 82       11p14.1d      BDNF          7         0.9728164
## 4       12q24.33d  KIAA0692          9         0.9714923
## 48      16p13.11b LOC339047          7         0.9696381
## 87      16p13.11b LOC339047          7         0.9692033
## 6       12p13.31a     DDX12         10         0.9685507
## 34       11p14.1d      BDNF          7         0.9681280
## 30      12p13.31a     DDX12         10         0.9657570
## 1       12p13.31a     DDX12         10         0.9617268
## 3               NA LOC653086          7         0.9575042
## 47        5q35.2d LOC202134          7         0.9568036
## 46       11p14.1d      BDNF          7         0.9289947
## 29        5q35.2d LOC202134          7         0.9285395
## 25      12q24.33d  KIAA0692          9         0.8823950
## 54 Xp21.2a-p21.1d       DMD          7         0.7956211
## 9         8q24.3g     PLEC1          8         0.7882088
## 39        5q35.2d LOC202134          7         0.3998123
## 21          4q23b     ADH1A          1         0.1841845
## 58       17q24.2c     ABCA8          1         0.1539030
## 32       14q24.3b       FOS          1         0.1197627
## 79      19q13.32a      FOSB          1         0.1190193
## 23       17q21.2b     KRT19          1         0.1159735
```

The above table gives the protein products, the ontology function, the fold change of the median values of UL/nonUL, gene symbol, sequence of CNV, and cytoband location. The protein products can be found at genecards.org by entering the ID for the protein product into the search bar. A quick scan of a few of the protein products in genecards.org gave the following descriptions. The first listed protein NP_061159.1 says it is a colon cancer secreted protein. Many of the above CNVs are listed as proteins involved in the extracellular matrix like DMD and CTNNB1. There are also various neurological and synapses diseases associated with those proteins.

The site genecards.org has very useful properties in analyzing gene expression data from this research. If you are a member, you can download the network genes involved in diseases you query and compare to how the genes in certain tissues compare to those genes. Three out of the seven CNVs for CTNNB1 are in the top fold change median values in the ratio of UL/nonUL samples.

```
write.csv(KG4, 'keyGenes_topMedFCs.csv', row.names=FALSE)
```

Lets make a machine learning data set to test various algorithms on predicting if the sample is a UL or not. We will use the samples in this set, plus add in some microarray samples that have been studied by me elsewhere using this set of genes and sequences if available in any of the microarray studies.

Lets isolate those genes that are in our key genes of top picks for UL targets and combine the UL and nonUL sample information to those genes and sequences without the stats.

```
keyTargets <- KG4[,c(1,4)]

ULs <- UL[,c(2,10:29)]
colnames(ULs)[2:21] <- paste('UL', colnames(ULs)[2:21], sep='_')

nonULs <- nonUL[,c(2,10:27)]
colnames(nonULs)[2:19] <- paste('nonUL', colnames(nonULs)[2:19], sep='_')

keyULs <- merge(keyTargets, ULs, by.x='SEQUENCE', by.y='SEQUENCE')
keys <- merge(keyULs, nonULs, by.x='SEQUENCE', by.y='SEQUENCE')

write.csv(keys,'keyGeneTargetsCNVs.csv',row.names=FALSE)
```

Lets create the matrix for machine learning.

```
keys_ml <- as.data.frame(t(keys))
colnames(keys_ml) <- paste(keys$Symbol,keys$SEQUENCE,sep='_')
keys_ml1 <- keys_ml[-(1:2),]
keys_ml1$Type <-
as.factor(c(rep('UL',length(grep('^UL_',row.names(keys_ml1)))),
                    rep('nonUL',length(grep('^nonUL_',row.names(keys_ml1))))))
keys_ml2 <- keys_ml1[,c(88,1:87)]
```

```
write.csv(keys_ml2, 'ml_ready_UL_classes.csv',row.names=TRUE)
```

Now, lets pull in the other data sets that are from the microarray samples and see if we can get the genes and sequences that correspond to our key genes above in identifying a sample as UL or not with predictive analytics.

There is one study of the other studies that has Sequence, Gene symbol and a few UL and nonUL microarray samples to compare to this above beadchip UL and nonUL set. The GEO series ID is GSE68295 with the GEO platform of GPL6480. The files are 27 MB each in file size.

```
setwd('./microArray UL')

non <- read.csv('nonUL_GSE68295_GPL6480_table.csv', sep=',',
                header=T, na.strings=c('',' '))
uls <- read.csv('UL_GSE68295_GPL6480_table.csv', sep=',',
                header=T, na.strings=c('',' '))
setwd('../')
```

Keep only the needed columns.

```
uls_array <- uls[,c(8,18:21)]
colnames(uls_array)[3:5] <- paste('UL', colnames(uls_array)[3:5], sep='_')

non_array <- non[,c(8,18:21)]
colnames(non_array)[3:5] <- paste('nonUL', colnames(non_array)[3:5], sep='_')
```

The sequences don't align or match any in the microarrays with the beadchip UL samples.

```
uls_array0 <- merge(keyTargets, uls_array, by.x='SEQUENCE', by.y='SEQUENCE')
ulsArray0 <- uls_array0[,-2]
```

Match by gene symbol between the microarray and beadchip UL samples.

```
uls_array1 <- merge(keyTargets, uls_array, by.x='Symbol', by.y='GENE_SYMBOL')
ulsArray <- uls_array1[,-2]
```

The sequences don't align between the arrays and beadchip samples for nonULs.

```
non_array0 <- merge(keyTargets, non_array, by.x='SEQUENCE', by.y='SEQUENCE')
nonArray0 <- non_array0[,-(1:2)]
```

Match by Gene symbol between the microarray and beadchip samples of nonULs.

```
non_array1 <- merge(keyTargets, non_array, by.x='Symbol', by.y='GENE_SYMBOL')
nonArray <- non_array1[,-(1:2)]
```

Combine the UL and nonUL samples of the microarrays into one dataset.

```
microarrays <- merge(ulsArray, nonArray, by.x='SEQUENCE.y',
by.y='SEQUENCE.y')
Marrays <- microarrays[!duplicated(microarrays$SEQUENCE),]
```

Since these two expression types can't be compared by sequence, they should be compared by gene. Lets combine them into a study by gene expression values.

```
keys1 <- keys[,-1]
keys2 <- keys1 %>% group_by(Symbol) %>%
  summarise_at(vars(as.vector(colnames(keys1)[2:39])), mean)

Marrays1 <- Marrays[,-1]
Marrays2 <- Marrays1 %>% group_by(Symbol) %>%
  summarise_at(vars(as.vector(colnames(Marrays1)[2:7])), mean)

beadArrays <- merge(keys2, Marrays2, by.x='Symbol', by.y='Symbol')
```

There are only 12 genes in common among these combined samples of microarray and beadchip UL and nonUL samples.

```
beadArrays_ML <- as.data.frame(t(beadArrays))
colnames(beadArrays_ML) <- beadArrays$Symbol
beadArrays_ML2 <- beadArrays_ML[-1,]
beadArrays_ML2$Type <- as.factor(c(rep('UL_bead',20), rep('nonUL_bead',18),
                         rep('UL_array',3), rep('nonUL_array',3)))
beadArrays_ML3 <- beadArrays_ML2[,c(13,1:12)]

UL_seq_ML <- keys_ml2
UL_gene_ML <- beadArrays_ML3
```

There are two datasets to use for machine learning. The first is our beadchip samples of 88 sequences and 38 samples of 20 UL and 18 nonUL in the **UL_seq_ML** data set. The second dataset for machine learning is the mixed microarray and beadchip samples of UL and nonUL by gene in the **UL_gene_ML** data set, because there were no common sequence or copy number variants of the gene sequences between the beadchip and microarray sets of UL and nonUL samples.

The libraries were installed earlier.

```
set.seed(02242020)
```

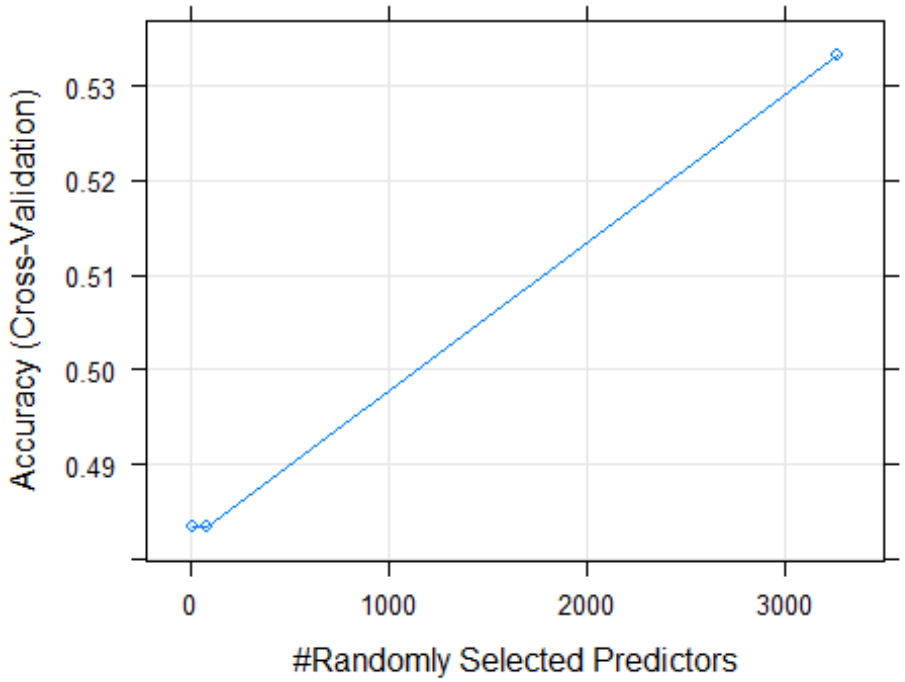
Create a partition of the data with a 70/30 split into training/testing sets of the first data set with two classes of UL or nonUL and 88 features of genes with their CNVs.

```
inTrain <- createDataPartition(y=UL_seq_ML$Type, p=0.7, list=FALSE)

trainingSet <- UL_seq_ML[inTrain,]
testingSet <- UL_seq_ML[-inTrain,]
```

RandomForest, cross-validation (cv) = 5

```
rfMod <- train(Type~., method='rf', data=(trainingSet),
               trControl=trainControl(method='cv'), number=5)
plot(rfMod)
```



Run predictions on the testing set

```
predRF <- predict(rfMod, testingSet)

predDF <- data.frame(predRF, type=testingSet$Type)
predDF

##    predRF  type
## 1      UL    UL
## 2      UL    UL
## 3      UL    UL
## 4      UL    UL
## 5      UL    UL
## 6      UL    UL
## 7      UL nonUL
## 8      UL nonUL
## 9      UL nonUL
## 10     UL nonUL
## 11     UL nonUL

sum <- sum(predRF==testingSet$Type)
length <- length(testingSet$Type)
```

```
accuracy_rfMod <- (sum/length)
accuracy_rfMod

## [1] 0.5454545

results <- c(round(accuracy_rfMod,2), round(100,2))
results <- as.factor(results)
results <- t(data.frame(results))

colnames(results) <- colnames(predDF)
Results <- rbind(predDF, results)
Results

##         predRF  type
## 1           UL    UL
## 2           UL    UL
## 3           UL    UL
## 4           UL    UL
## 5           UL    UL
## 6           UL    UL
## 7           UL nonUL
## 8           UL nonUL
## 9           UL nonUL
## 10          UL nonUL
## 11          UL nonUL
## results   0.55   100
```

The above shows that using the genes and the CNV of each gene totalling 88 features, makes a poor data set of results with only 27 observations to train and 11 to test on 2 classes of UL or non-UL.

What if we used random forest to only predict by gene in the first beadchip type data set? We can use the transpose of the keys2 data set made earlier when combining to make the 2nd ML dataset.

```
keys_t <- as.data.frame(t(keys2))
colnames(keys_t) <- keys2$Symbol
keys_t1 <- keys_t[-1,]
keys_t1$Type <- keys_ml2$Type
keys_ML <- keys_t1[,c(21,1:20)]
```

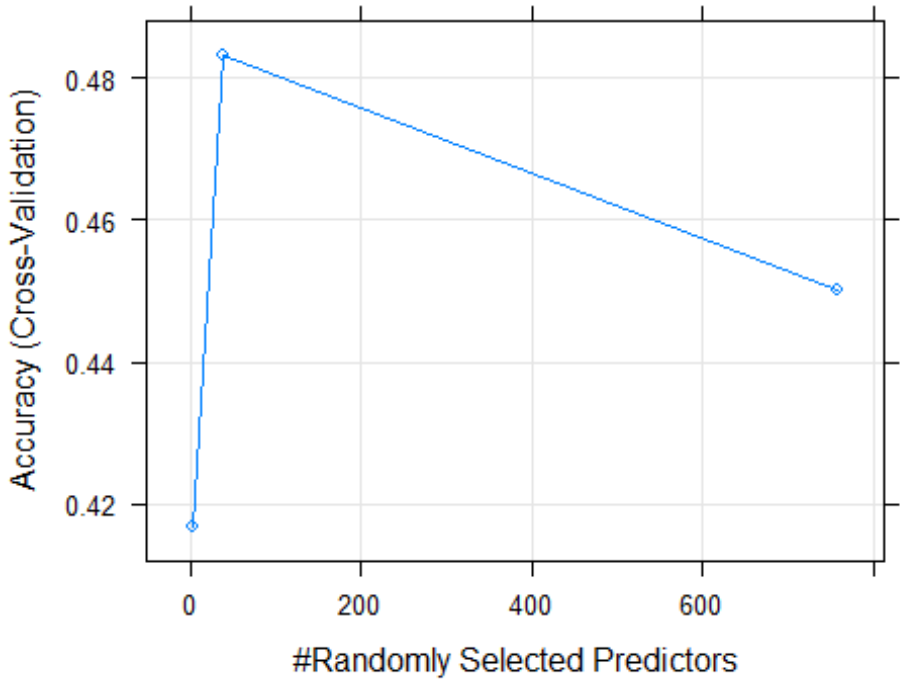
Now we will use our new data set based on the beadchip genes and not the CNVs of those genes, in the keys_ML data set to predict with RandomForest.

```
inTrain <- createDataPartition(y=keys_ML$Type, p=0.7, list=FALSE)

trainingSet <- keys_ML[inTrain,]
testingSet <- keys_ML[-inTrain,]
```

RandomForest, cross-validation (cv) = 5

```
rfMod <- train(Type~., method='rf', data=(trainingSet),
               trControl=trainControl(method='cv'), number=5)
plot(rfMod)
```



Run predictions on the testing set

```
predRF <- predict(rfMod, testingSet)

predDF <- data.frame(predRF, type=testingSet$Type)
predDF
```

```
##    predRF  type
## 1      UL    UL
## 2      UL    UL
## 3      UL    UL
## 4      UL    UL
## 5      UL    UL
## 6      UL    UL
## 7      UL nonUL
## 8      UL nonUL
## 9      UL nonUL
## 10     UL nonUL
## 11     UL nonUL
```

```
sum <- sum(predRF==testingSet$Type)
length <- length(testingSet$Type)
```

```
accuracy_rfMod <- (sum/length)
accuracy_rfMod

## [1] 0.5454545

results <- c(round(accuracy_rfMod,2), round(100,2))
results <- as.factor(results)
results <- t(data.frame(results))

colnames(results) <- colnames(predDF)
Results <- rbind(predDF, results)
Results

##         predRF  type
## 1           UL    UL
## 2           UL    UL
## 3           UL    UL
## 4           UL    UL
## 5           UL    UL
## 6           UL    UL
## 7           UL nonUL
## 8           UL nonUL
## 9           UL nonUL
## 10          UL nonUL
## 11          UL nonUL
## results   0.55   100
```

How about with the KNN algorithm.

```
knnMod <- train(Type ~ .,
                method='knn', preProcess=c('center','scale'),
                tuneLength=10, trControl=trainControl(method='cv'),
data=trainingSet)

## Warning in preProcess.default(thresh = 0.95, k = 5, freqCut = 19,
uniqueCut =
## 10, : These variables have zero variances: ABCA8 88.61720, ABCA8 77.21540,
ABCA8
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## KIAA1199 1748.59090, KIAA1199 200.75370, KIAA11991502.76260,
KIAA11992212.23820,
## KIAA11992841.15650, KIAA11993452.39830, KIAA11994272.82880,
KIAA11996548.81600,
## KIAA1199KIAA1199, KRT19 110.76330, KRT19 49.10000, KRT19 57.40000, KRT19
## 61.56670, KRT19 90.88670, KRT19 168.43680, KRT19 173.69500, KRT19
229.34340,
## KRT19 232.75320, KRT19 515.85060, KRT19 656.00690, KRT19 773.91250, KRT19
## 825.54400, KRT191091.95250, KRT19KRT19, LOC202134 92.87717, LOC202134
## 80.73967, LOC202134 80.93756, LOC202134 84.86834, LOC202134 86.33883,
LOC202134
## 91.90153, LOC202134 97.46411, LOC202134 97.86973, LOC202134 99.14780,
LOC202134
## 99.72284, LOC202134 100.08753, LOC202134 100.62161, LOC202134 125.11937,
## LOC202134 153.49023, LOC202134LOC202134, LOC23117 58.45052, LOC23117
56.78304,
## LOC23117 56.92304, LOC23117 57.00729, LOC23117 57.58250, LOC23117
57.63400,
## LOC23117 57.87063, LOC23117 57.87225, LOC23117 58.25216, LOC23117
59.49375,
## LOC23117 59.70365, LOC23117 60.84021, LOC23117 61.10820, LOC23117
64.34166,
## LOC23117LOC23117, LOC339047 52.66400, LOC339047 53.31190, LOC339047
53.39899,
## LOC339047 54.63439, LOC339047 54.90714, LOC339047 54.96286, LOC339047
55.02857,
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## LOC339047 55.83333, LOC339047 56.06596, LOC339047 56.25000, LOC339047
56.88054,
## LOC339047 57.71951, LOC339047 58.38786, LOC339047LOC339047, LOC653086
909.83774,
## LOC653086 390.88886, LOC653086 417.10514, LOC653086 648.07329, LOC653086
## 660.33487, LOC653086 772.56214, LOC653086 825.04971, LOC653086 852.31269,
## LOC653086 870.00770, LOC653086 897.15661, LOC653086 925.40631, LOC653086
## 935.03846, LOC6530861065.10449, LOC6530861122.18651, LOC653086LOC653086,
## MMP11 75.97500, MMP11 84.25000, MMP11 234.30210, MMP11 235.96740, MMP11
## 318.05450, MMP11 375.42970, MMP11 443.38950, MMP111338.71330,
MMP1115286.34750,
## MMP113590.97470, MMP113713.31130, MMP114141.86420, MMP115414.01170,
## MMP118858.71940, MMP11MMP11, PENK 54.20000, PENK 59.48000, PENK 60.30000,
PENK
## 61.60000, PENK 78.26670, PENK 99.13530, PENK 166.37800, PENK 183.02000,
PENK
## 214.79190, PENK 255.79600, PENK 628.35690, PENK 699.56800,
PENK26134.71050,
## PENK2779.81960, PENKPENK, PLEC1 63.21976, PLEC1 68.78959, PLEC1 69.57295,
PLEC1
## 70.89612, PLEC1 71.50187, PLEC1 76.05800, PLEC1 76.19250, PLEC1 78.75296,
PLEC1
## 79.68368, PLEC1 85.84841, PLEC1 94.55256, PLEC1 109.94919, PLEC1
116.72813,
## PLEC1PLEC1

## Warning in preProcess.default(thresh = 0.95, k = 5, freqCut = 19,
uniqueCut =
## 10, : These variables have zero variances: ABCA8 88.61720, ABCA8 77.21540,
ABCA8
## 92.13060, ABCA8 96.40000, ABCA8 97.56250, ABCA8 110.85790, ABCA8
125.96360,
## ABCA8 158.93000, ABCA8 206.22750, ABCA8 521.93650, ABCA8 576.07270, ABCA8
## 586.46630, ABCA8 703.32080, ABCA8 761.93730, ABCA8ABCA8, ACTC 2077.75710,
ACTC
## 209.25830, ACTC 216.30000, ACTC 256.80780, ACTC 277.35920, ACTC 320.94930,
## ACTC 350.72790, ACTC 830.23350, ACTC1205.77090, ACTC5434.41450,
ACTC6205.93360,
## ACTC6894.49490, ACTC7325.32550, ACTC8327.85490, ACTCACTC, ADH1A 153.07270,
## ADH1A 275.28830, ADH1A 320.97780, ADH1A 517.43540, ADH1A 640.42100, ADH1A
## 660.45380, ADH1A 873.77740, ADH1A1157.79510, ADH1A1936.02330,
ADH1A1997.93640,
## ADH1A1998.15530, ADH1A3085.55830, ADH1A3647.80130, ADH1AADH1A, BDNF
73.08220,
## BDNF 58.65697, BDNF 59.53214, BDNF 61.17047, BDNF 61.44807, BDNF 62.29957,
## BDNF 62.38321, BDNF 64.23757, BDNF 64.47564, BDNF 64.79746, BDNF 65.37834,
## BDNF 69.25817, BDNF 71.83247, BDNF 88.13646, BDNFBDNF, CTNNB1 687.48347,
## CTNNB1 482.59696, CTNNB1 492.99827, CTNNB1 519.50586, CTNNB1 538.13661,
## CTNNB1 599.45751, CTNNB1 626.21914, CTNNB1 656.16527, CTNNB1 759.35247,
## CTNNB1 762.08883, CTNNB1 993.12030, CTNNB11154.33303, CTNNB11178.59467,
```

```
## CTNNB11179.90051, CTNNB1CTNNB1, DDX12 53.43858, DDX12 53.20944, DDX12
54.36233,
## DDX12 54.58100, DDX12 54.73183, DDX12 54.94688, DDX12 55.16945, DDX12
55.32804,
## DDX12 55.46834, DDX12 55.81690, DDX12 55.97067, DDX12 56.22057, DDX12
58.59496,
## DDX12 61.05796, DDX12DDX12, DLK1 195.52710, DLK1 48.50000, DLK1 48.70000,
DLK1
## 51.00000, DLK1 51.50000, DLK1 52.45000, DLK1 58.50000, DLK1 146.61360,
DLK1
## 175.13040, DLK1 272.58790, DLK1 384.38110, DLK1 475.95320, DLK1 921.19970,
## DLK12828.10710, DLK1DLK1, DMD 103.29663, DMD 60.21667, DMD 87.96567, DMD
## 98.12929, DMD 103.15579, DMD 134.62189, DMD 137.26089, DMD 137.43906, DMD
## 145.11183, DMD 148.46514, DMD 151.27870, DMD 152.12489, DMD 186.47406, DMD
## 219.38810, DMDDMD, FOS 386.81900, FOS 577.79010, FOS 588.20420, FOS
845.98350,
## FOS1216.05550, FOS1476.46510, FOS2288.23880, FOS3473.41090, FOS3514.50890,
## FOS3868.98980, FOS5620.43240, FOS6842.36240, FOS9423.57470, FOSFOS, FOSB
## 57.90000, FOSB 59.50000, FOSB 61.90000, FOSB 64.62220, FOSB 72.47000, FOSB
## 95.95830, FOSB 135.03820, FOSB 207.68810, FOSB 321.31970, FOSB 601.59350,
## FOSB 803.93670, FOSB1162.21760, FOSB1211.49910, FOSBFOSB, KIAA0692
58.21603,
## KIAA0692 52.08757, KIAA0692 56.17667, KIAA0692 56.32444, KIAA0692
58.06852,
## KIAA0692 59.19570, KIAA0692 60.28206, KIAA0692 63.07222, KIAA0692
64.16567,
## KIAA0692 65.04341, KIAA0692 66.84581, KIAA0692 67.24871, KIAA0692
68.57124,
## KIAA0692 69.12237, KIAA0692KIAA0692, KIAA1199 63.10000, KIAA1199 69.80000,
## KIAA1199 93.08750, KIAA1199 94.10810, KIAA1199 114.87180, KIAA1199
156.83750,
## KIAA1199 1748.59090, KIAA1199 200.75370, KIAA11991502.76260,
KIAA11992212.23820,
## KIAA11992841.15650, KIAA11993452.39830, KIAA11994272.82880,
KIAA11996548.81600,
## KIAA1199KIAA1199, KRT19 110.76330, KRT19 49.10000, KRT19 57.40000, KRT19
## 61.56670, KRT19 90.88670, KRT19 168.43680, KRT19 173.69500, KRT19
229.34340,
## KRT19 232.75320, KRT19 515.85060, KRT19 656.00690, KRT19 773.91250, KRT19
## 825.54400, KRT191091.95250, KRT19KRT19, LOC202134 92.87717, LOC202134
## 80.73967, LOC202134 80.93756, LOC202134 84.86834, LOC202134 86.33883,
LOC202134
## 91.90153, LOC202134 97.46411, LOC202134 97.86973, LOC202134 99.14780,
LOC202134
## 99.72284, LOC202134 100.08753, LOC202134 100.62161, LOC202134 125.11937,
## LOC202134 153.49023, LOC202134LOC202134, LOC23117 58.45052, LOC23117
56.78304,
## LOC23117 56.92304, LOC23117 57.00729, LOC23117 57.58250, LOC23117
57.63400,
## LOC23117 57.87063, LOC23117 57.87225, LOC23117 58.25216, LOC23117
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59.49375,
## LOC23117 59.70365, LOC23117 60.84021, LOC23117 61.10820, LOC23117
64.34166,
## LOC23117LOC23117, LOC339047 52.66400, LOC339047 53.31190, LOC339047
53.39899,
## LOC339047 54.63439, LOC339047 54.90714, LOC339047 54.96286, LOC339047
55.02857,
## LOC339047 55.83333, LOC339047 56.06596, LOC339047 56.25000, LOC339047
56.88054,
## LOC339047 57.71951, LOC339047 58.38786, LOC339047LOC339047, LOC653086
909.83774,
## LOC653086 390.88886, LOC653086 417.10514, LOC653086 648.07329, LOC653086
## 660.33487, LOC653086 772.56214, LOC653086 825.04971, LOC653086 852.31269,
## LOC653086 870.00770, LOC653086 897.15661, LOC653086 925.40631, LOC653086
## 935.03846, LOC6530861065.10449, LOC6530861122.18651, LOC653086LOC653086,
## MMP11 75.97500, MMP11 84.25000, MMP11 234.30210, MMP11 235.96740, MMP11
## 318.05450, MMP11 375.42970, MMP11 443.38950, MMP111338.71330,
MMP1115286.34750,
## MMP113590.97470, MMP113713.31130, MMP114141.86420, MMP115414.01170,
## MMP118858.71940, MMP11MMP11, PENK 54.20000, PENK 59.48000, PENK 60.30000,
PENK
## 61.60000, PENK 78.26670, PENK 99.13530, PENK 166.37800, PENK 183.02000,
PENK
## 214.79190, PENK 255.79600, PENK 628.35690, PENK 699.56800,
PENK26134.71050,
## PENK2779.81960, PENKPENK, PLEC1 63.21976, PLEC1 68.78959, PLEC1 69.57295,
PLEC1
## 70.89612, PLEC1 71.50187, PLEC1 76.05800, PLEC1 76.19250, PLEC1 78.75296,
PLEC1
## 79.68368, PLEC1 85.84841, PLEC1 94.55256, PLEC1 109.94919, PLEC1
116.72813,
## PLEC1PLEC1

## Warning in preProcess.default(thresh = 0.95, k = 5, freqCut = 19,
uniqueCut =
## 10, : These variables have zero variances: ABCA8 88.61720, ABCA8 77.21540,
ABCA8
## 96.40000, ABCA8 110.85790, ABCA8 125.96360, ABCA8 158.93000, ABCA8
206.22750,
## ABCA8 576.07270, ABCA8 586.46630, ABCA8 703.32080, ABCA8 761.93730,
ABCA8ABCA8,
## ACTC 2077.75710, ACTC 216.30000, ACTC 277.35920, ACTC 320.94930, ACTC
350.72790,
## ACTC 830.23350, ACTC1205.77090, ACTC5434.41450, ACTC6205.93360,
ACTC7325.32550,
## ACTC8327.85490, ACTCACTC, ADH1A 153.07270, ADH1A 275.28830, ADH1A
517.43540,
## ADH1A 660.45380, ADH1A 873.77740, ADH1A1157.79510, ADH1A1936.02330,
## ADH1A1997.93640, ADH1A1998.15530, ADH1A3647.80130, ADH1AADH1A, BDNF
73.08220,
```
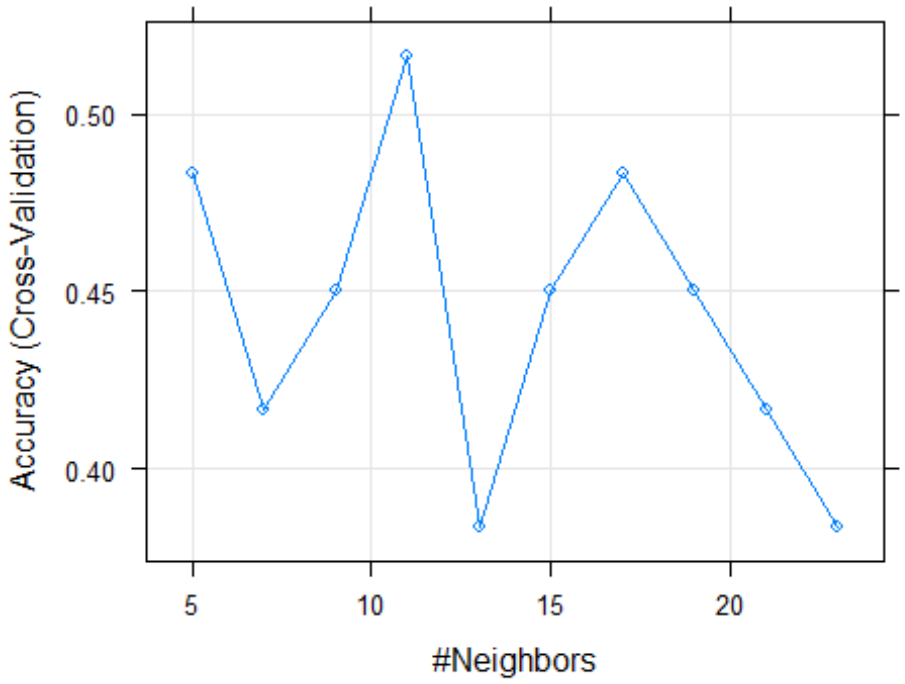

```
## BDNF 58.65697, BDNF 59.53214, BDNF 61.44807, BDNF 62.29957, BDNF 62.38321,
BDNF
## 64.23757, BDNF 65.37834, BDNF 69.25817, BDNF 71.83247, BDNF 88.13646,
BDNFBDNF,
## CTNNB1 687.48347, CTNNB1 492.99827, CTNNB1 519.50586, CTNNB1 538.13661,
## CTNNB1 599.45751, CTNNB1 656.16527, CTNNB1 759.35247, CTNNB1 762.08883,
CTNNB1
## 993.12030, CTNNB11178.59467, CTNNB11179.90051, CTNNB1CTNNB1, DDX12
53.43858,
## DDX12 53.20944, DDX12 54.36233, DDX12 54.58100, DDX12 54.73183, DDX12
54.94688,
## DDX12 55.46834, DDX12 55.81690, DDX12 56.22057, DDX12 58.59496, DDX12
61.05796,
## DDX12DDX12, DLK1 195.52710, DLK1 48.50000, DLK1 48.70000, DLK1 51.00000,
DLK1
## 52.45000, DLK1 58.50000, DLK1 146.61360, DLK1 175.13040, DLK1 272.58790,
DLK1
## 475.95320, DLK1 921.19970, DLK1DLK1, DMD 103.29663, DMD 60.21667, DMD
98.12929,
## DMD 103.15579, DMD 134.62189, DMD 137.26089, DMD 137.43906, DMD 148.46514,
DMD
## 152.12489, DMD 186.47406, DMD 219.38810, DMDDMD, FOS 386.81900, FOS
588.20420,
## FOS1216.05550, FOS1476.46510, FOS2288.23880, FOS3473.41090, FOS3514.50890,
## FOS3868.98980, FOS5620.43240, FOS9423.57470, FOSFOS, FOSB 59.50000, FOSB
## 64.62220, FOSB 72.47000, FOSB 95.95830, FOSB 135.03820, FOSB 207.68810,
FOSB
## 601.59350, FOSB 803.93670, FOSB1162.21760, FOSB1211.49910, FOSBFOSB,
KIAA0692
## 58.21603, KIAA0692 52.08757, KIAA0692 58.06852, KIAA0692 59.19570,
KIAA0692
## 60.28206, KIAA0692 63.07222, KIAA0692 64.16567, KIAA0692 65.04341,
KIAA0692
## 67.24871, KIAA0692 68.57124, KIAA0692 69.12237, KIAA0692KIAA0692, KIAA1199
## 69.80000, KIAA1199 93.08750, KIAA1199 94.10810, KIAA1199 114.87180,
KIAA1199
## 1748.59090, KIAA1199 200.75370, KIAA11991502.76260, KIAA11992212.23820,
## KIAA11992841.15650, KIAA11993452.39830, KIAA11994272.82880,
KIAA1199KIAA1199,
## KRT19 110.76330, KRT19 57.40000, KRT19 61.56670, KRT19 90.88670, KRT19
## 168.43680, KRT19 229.34340, KRT19 232.75320, KRT19 656.00690, KRT19
773.91250,
## KRT19 825.54400, KRT191091.95250, KRT19KRT19, LOC202134 92.87717,
LOC202134
## 80.73967, LOC202134 80.93756, LOC202134 86.33883, LOC202134 91.90153,
LOC202134
## 99.14780, LOC202134 99.72284, LOC202134 100.08753, LOC202134 100.62161,
## LOC202134 125.11937, LOC202134 153.49023, LOC202134LOC202134, LOC23117
## 58.45052, LOC23117 56.78304, LOC23117 57.00729, LOC23117 57.58250,
LOC23117
```

```
## 57.63400, LOC23117 57.87063, LOC23117 57.87225, LOC23117 58.25216,
LOC23117
## 59.70365, LOC23117 61.10820, LOC23117 64.34166, LOC23117LOC23117,
LOC339047
## 52.66400, LOC339047 53.39899, LOC339047 54.63439, LOC339047 54.96286,
LOC339047
## 55.83333, LOC339047 56.06596, LOC339047 56.25000, LOC339047 56.88054,
LOC339047
## 57.71951, LOC339047 58.38786, LOC339047LOC339047, LOC653086 909.83774,
LOC653086
## 390.88886, LOC653086 648.07329, LOC653086 660.33487, LOC653086 772.56214,
## LOC653086 825.04971, LOC653086 852.31269, LOC653086 870.00770, LOC653086
## 897.15661, LOC6530861065.10449, LOC6530861122.18651, LOC653086LOC653086,
## MMP11 75.97500, MMP11 84.25000, MMP11 318.05450, MMP11 375.42970, MMP11
## 443.38950, MMP111338.71330, MMP1115286.34750, MMP113590.97470,
MMP113713.31130,
## MMP115414.01170, MMP118858.71940, MMP11MMP11, PENK 54.20000, PENK
59.48000,
## PENK 60.30000, PENK 61.60000, PENK 99.13530, PENK 166.37800, PENK
183.02000,
## PENK 214.79190, PENK 628.35690, PENK 699.56800, PENK26134.71050, PENKPENK,
## PLEC1 63.21976, PLEC1 68.78959, PLEC1 69.57295, PLEC1 71.50187, PLEC1
76.19250,
## PLEC1 78.75296, PLEC1 79.68368, PLEC1 85.84841, PLEC1 94.55256, PLEC1
116.72813,
## PLEC1PLEC1

plot(knnMod)
```



The accuracy seems to be better between 8 and 9 neighbors for classification from what the above plot is displaying.

```
rpartMod <- train(Type ~ ., method='rpart', tuneLength=7, data=trainingSet)

glmMod <- train(Type ~ .,
                method='glm', data=trainingSet)

predKNN <- predict(knnMod, testingSet)
predRPART <- predict(rpartMod, testingSet)
predGLM <- predict(glmMod, testingSet)

length=length(testingSet$Type)

sumKNN <- sum(predKNN==testingSet$Type)
sumRPart <- sum(predRPART==testingSet$Type)
sumGLM <- sum(predGLM==testingSet$Type)

accuracy_KNN <- sumKNN/length
accuracy_RPART <- sumRPart/length
accuracy_GLM <- sumGLM/length

predDF2 <- data.frame(predRF,predKNN,predRPART,predGLM,
                      TYPE=testingSet$Type)
colnames(predDF2) <- c('RandomForest','KNN','Rpart','GLM','TrueValue')

results <- c(round(accuracy_rfMod,2),
```

```
              round(accuracy_KNN,2),
              round(accuracy_RPART,2),
              round(accuracy_GLM,2),
              round(100,2))

results <- as.factor(results)
results <- t(data.frame(results))
colnames(results) <- c('RandomForest','KNN','Rpart','GLM','TrueValue')
Results <- rbind(predDF2, results)
Results
```

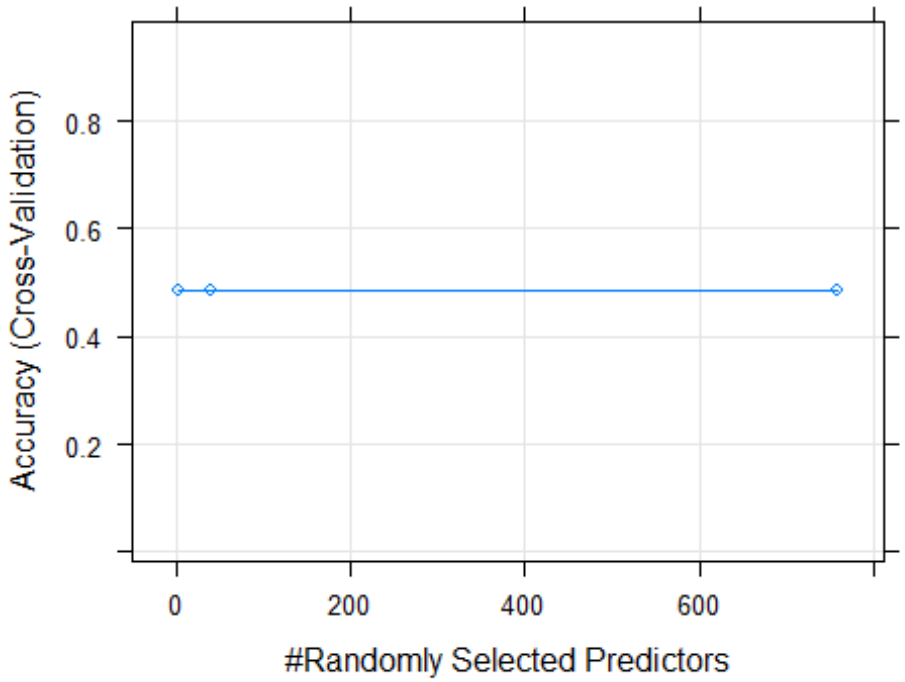
```
##         RandomForest  KNN Rpart  GLM TrueValue
## 1                 UL   UL    UL   UL        UL
## 2                 UL   UL    UL   UL        UL
## 3                 UL   UL    UL   UL        UL
## 4                 UL   UL    UL   UL        UL
## 5                 UL   UL    UL   UL        UL
## 6                 UL   UL    UL   UL        UL
## 7                 UL   UL    UL   UL     nonUL
## 8                 UL   UL    UL   UL     nonUL
## 9                 UL   UL    UL   UL     nonUL
## 10                UL   UL    UL   UL     nonUL
## 11                UL   UL    UL   UL     nonUL
## results         0.55 0.55  0.55 0.55       100
```

As far as the algorithms used above go, the prediction accuracy is not good at all. This could be because there aren't enough samples. This could also be because there are a lot of features and not enough samples. We could try just predicting the sample type using less features, like the genes with the highest fold change values. Those genes were KIAA1199, ACTC, MMP11, FOSB, KRT19, and FOSB with the three most over/under expressed in fold change mean values.

```
keys_ML_b <- keys_ML[,c(1,3,10,11,13,19)]

inTrain <- createDataPartition(y=keys_ML_b$Type, p=0.7, list=FALSE)

trainingSet <- keys_ML[inTrain,]
testingSet <- keys_ML[-inTrain,]
```

RandomForest, cross-validation (cv) = 5

```
rfMod <- train(Type~., method='rf', data=(trainingSet),
               trControl=trainControl(method='cv'), number=5)
plot(rfMod)
```



Run predictions on the testing set

```
predRF <- predict(rfMod, testingSet)

predDF <- data.frame(predRF, type=testingSet$Type)
predDF
```

```
##    predRF  type
## 1      UL    UL
## 2      UL    UL
## 3      UL    UL
## 4      UL    UL
## 5      UL    UL
## 6      UL    UL
## 7      UL nonUL
## 8      UL nonUL
## 9      UL nonUL
## 10     UL nonUL
## 11     UL nonUL
```

```
sum <- sum(predRF==testingSet$Type)
length <- length(testingSet$Type)
accuracy_rfMod <- (sum/length)
accuracy_rfMod
```

```
## [1] 0.5454545
```

```
results <- c(round(accuracy_rfMod,2), round(100,2))
results <- as.factor(results)
results <- t(data.frame(results))

colnames(results) <- colnames(predDF)
Results <- rbind(predDF, results)
Results
```

```
##         predRF  type
## 1           UL    UL
## 2           UL    UL
## 3           UL    UL
## 4           UL    UL
## 5           UL    UL
## 6           UL    UL
## 7           UL nonUL
## 8           UL nonUL
## 9           UL nonUL
## 10          UL nonUL
## 11          UL nonUL
## results   0.55   100
```

How about with the KNN algorithm.

```
knnMod <- train(Type ~ .,
                method='knn', preProcess=c('center','scale'),
                tuneLength=10, trControl=trainControl(method='cv'),
data=trainingSet)
```

```
## Warning in preProcess.default(thresh = 0.95, k = 5, freqCut = 19,
uniqueCut
## = 10, : These variables have zero variances: ABCA8 155.80000, ABCA8
56.05000,
## ABCA8 681.04640, ABCA8 92.13060, ABCA8 96.40000, ABCA8 125.96360, ABCA8
## 132.38820, ABCA8 292.93330, ABCA8 455.64560, ABCA8 562.68870, ABCA8
576.07270,
## ABCA8 830.87020, ABCA81493.23800, ABCA8ABCA8, ACTC 478.79120, ACTC
1452.70000,
## ACTC 169.41300, ACTC 222.50000, ACTC 320.94930, ACTC 350.72790, ACTC
622.15940,
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## DLK1 384.38110, DLK1 475.95320, DLK14126.77500, DLK1DLK1, DMD 132.51420,
## DMD 173.72703, DMD 60.21667, DMD 87.96567, DMD 100.27049, DMD 103.15579,
DMD
## 134.31964, DMD 136.18699, DMD 137.43906, DMD 148.46514, DMD 152.12489, DMD
## 157.94423, DMD 181.68309, DMD 223.08834, DMDDMD, FOS 621.78120, FOS
220.68100,
## FOS 386.81900, FOS 417.75610, FOS 577.79010, FOS 612.89650, FOS1216.05550,
## FOS17362.03170, FOS2995.18830, FOS3473.41090, FOS3514.50890,
FOS3868.98980,
## FOS8590.64340, FOS9819.01050, FOSFOS, FOSB 95.39480, FOSB 57.90000, FOSB
## 60.92000, FOSB 61.65000, FOSB 64.62220, FOSB 95.95830, FOSB 110.60000,
FOSB
## 207.68810, FOSB 258.97840, FOSB 4247.93770, FOSB 601.59350, FOSB
680.53840,
## FOSB1211.49910, FOSB7604.02210, FOSBFOSB, KIAA0692 62.88800, KIAA0692
67.28364,
## KIAA0692 53.62629, KIAA0692 58.06852, KIAA0692 59.19570, KIAA0692
60.28206,
## KIAA0692 64.27382, KIAA0692 64.75153, KIAA0692 64.89243, KIAA0692
65.02993,
## KIAA0692 65.04341, KIAA0692 66.84581, KIAA0692 67.47397, KIAA0692
69.12237,
## KIAA0692KIAA0692, KIAA1199 65.80000, KIAA1199 69.80000, KIAA1199 93.08750,
## KIAA1199 200.75370, KIAA1199 212.18080, KIAA1199 5039.24990,
KIAA11991798.94770,
## KIAA11992841.15650, KIAA11993915.91920, KIAA11994272.82880,
KIAA11994624.54100,
## KIAA11996160.16650, KIAA11996548.81600, KIAA1199KIAA1199, KRT19 45.60000,
KRT19
## 57.40000, KRT19 577.13950, KRT19 58.18000, KRT19 168.43680, KRT19
173.69500,
## KRT19 180.82220, KRT19 232.75320, KRT19 293.32220, KRT19 311.20200, KRT19
## 773.91250, KRT19 825.54400, KRT19 992.90020, KRT19KRT19, LOC202134
90.27716,
## LOC202134 116.28997, LOC202134 80.93756, LOC202134 81.09227, LOC202134
## 81.95797, LOC202134 91.90153, LOC202134 95.92870, LOC202134 97.86973,
LOC202134
## 99.14780, LOC202134 99.72284, LOC202134 103.06420, LOC202134 103.91630,
```

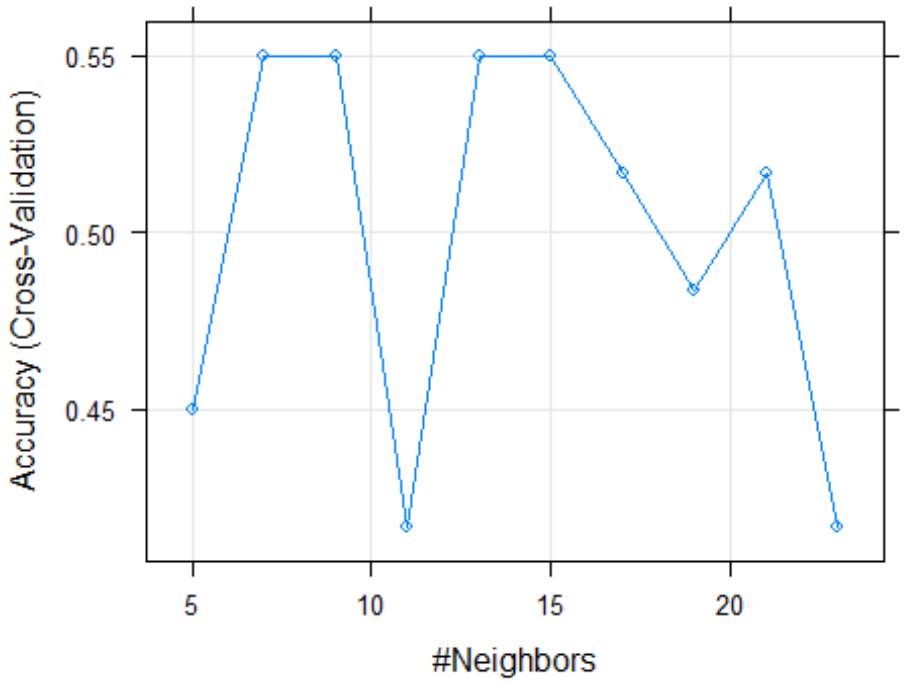
```
## LOC202134 109.43154, LOC202134 153.49023, LOC202134LOC202134, LOC23117
58.95793,
## LOC23117 63.41986, LOC23117 56.78304, LOC23117 56.92304, LOC23117
57.00729,
## LOC23117 57.14000, LOC23117 57.58250, LOC23117 57.87063, LOC23117
58.72856,
## LOC23117 59.18544, LOC23117 59.26586, LOC23117 59.70365, LOC23117
60.51843,
## LOC23117 66.15578, LOC23117LOC23117, LOC339047 54.32226, LOC339047
55.18711,
## LOC339047 53.39899, LOC339047 53.41786, LOC339047 54.22306, LOC339047
54.90714,
## LOC339047 54.96286, LOC339047 54.99321, LOC339047 55.83333, LOC339047
56.07547,
## LOC339047 57.69556, LOC339047 57.71951, LOC339047 58.38786, LOC339047
58.47381,
## LOC339047LOC339047, LOC653086 754.31924, LOC653086 841.71641, LOC653086
## 390.88886, LOC653086 477.83457, LOC653086 735.69129, LOC653086 764.01074,
## LOC653086 772.56214, LOC653086 825.04971, LOC653086 870.00770, LOC653086
## 897.15661, LOC653086 922.11347, LOC653086 925.40631, LOC6530861066.72823,
## LOC6530861203.33167, LOC653086LOC653086, MMP11 75.97500, MMP11 84.25000,
## MMP11 136.19640, MMP11 199.35870, MMP11 443.38950, MMP11 5787.43480, MMP11
## 669.80360, MMP111875.99000, MMP112082.50560, MMP113713.31130,
MMP114141.86420,
## MMP118300.29490, MMP118858.71940, MMP11MMP11, PENK 158.16150, PENK
248.20700,
## PENK 48.20000, PENK 54.20000, PENK 55.70000, PENK 59.48000, PENK 60.38330,
PENK
## 61.60000, PENK 89.07890, PENK 214.79190, PENK 219.76200, PENK 255.79600,
PENK
## 699.56800, PENK2788.93790, PENKPENK, PLEC1 117.83730, PLEC1 187.59291,
PLEC1
## 66.99375, PLEC1 68.78959, PLEC1 69.57295, PLEC1 70.30182, PLEC1 76.05800,
PLEC1
## 76.19250, PLEC1 79.68368, PLEC1 94.55256, PLEC1 94.90675, PLEC1 104.29716,
PLEC1
## 108.11144, PLEC1 137.53784, PLEC1PLEC1

## Warning in preProcess.default(thresh = 0.95, k = 5, freqCut = 19,
uniqueCut
## = 10, : These variables have zero variances: ABCA8 56.05000, ABCA8
681.04640,
## ABCA8 92.13060, ABCA8 96.40000, ABCA8 125.96360, ABCA8 132.38820, ABCA8
## 576.07270, ABCA8 830.87020, ABCA81493.23800, ABCA8ABCA8, ACTC 478.79120,
ACTC
## 169.41300, ACTC 320.94930, ACTC 350.72790, ACTC 622.15940,
ACTC13992.26470,
## ACTC4004.56310, ACTC5419.39300, ACTC6894.49490, ACTC7028.38050,
ACTC8327.85490,
## ACTCACTC, ADH1A 171.55770, ADH1A 54.35000, ADH1A 1797.60150, ADH1A
```

```
260.63550,
## ADH1A 275.28830, ADH1A 640.42100, ADH1A 873.77740, ADH1A1503.13120,
## ADH1A1944.23980, ADH1A1998.15530, ADH1A7727.82380, ADH1AADH1A, BDNF
66.79751,
## BDNF 68.67747, BDNF 59.53214, BDNF 61.34184, BDNF 62.38321, BDNF 62.63791,
BDNF
## 63.00849, BDNF 64.23757, BDNF 64.79746, BDNF 64.93227, BDNF 66.47571,
BDNFBDNF,
## CTNNB1 430.51867, CTNNB1 427.31916, CTNNB1 599.45751, CTNNB1 626.21914,
## CTNNB1 629.60446, CTNNB1 656.16527, CTNNB1 661.17213, CTNNB1 701.44551,
CTNNB1
## 736.31490, CTNNB1 762.08883, CTNNB1CTNNB1, DDX12 55.51556, DDX12 56.68058,
## DDX12 53.20944, DDX12 53.62324, DDX12 54.58100, DDX12 55.09119, DDX12
55.16945,
## DDX12 57.06467, DDX12 58.59496, DDX12 59.08089, DDX12 66.08112,
DDX12DDX12,
## DLK1 121.15160, DLK1 47.30000, DLK1 51.00000, DLK1 51.72500, DLK1
55.00000,
## DLK1 58.50000, DLK1 150.24510, DLK1 384.38110, DLK1 475.95320,
DLK14126.77500,
## DLK1DLK1, DMD 132.51420, DMD 173.72703, DMD 60.21667, DMD 87.96567, DMD
## 100.27049, DMD 103.15579, DMD 136.18699, DMD 152.12489, DMD 157.94423, DMD
## 181.68309, DMD 223.08834, DMDDMD, FOS 621.78120, FOS 220.68100, FOS
386.81900,
## FOS 417.75610, FOS 577.79010, FOS 612.89650, FOS17362.03170,
FOS3514.50890,
## FOS3868.98980, FOS8590.64340, FOS9819.01050, FOSFOS, FOSB 95.39480, FOSB
## 57.90000, FOSB 60.92000, FOSB 61.65000, FOSB 64.62220, FOSB 110.60000,
FOSB
## 207.68810, FOSB 4247.93770, FOSB 601.59350, FOSB 680.53840,
FOSB7604.02210,
## FOSBFOSB, KIAA0692 62.88800, KIAA0692 67.28364, KIAA0692 58.06852,
## KIAA0692 60.28206, KIAA0692 64.27382, KIAA0692 64.75153, KIAA0692
64.89243,
## KIAA0692 65.02993, KIAA0692 66.84581, KIAA0692 67.47397, KIAA0692
69.12237,
## KIAA0692KIAA0692, KIAA1199 65.80000, KIAA1199 93.08750, KIAA1199
200.75370,
## KIAA1199 212.18080, KIAA1199 5039.24990, KIAA11991798.94770,
KIAA11994272.82880,
## KIAA11994624.54100, KIAA11996160.16650, KIAA11996548.81600,
KIAA1199KIAA1199,
## KRT19 45.60000, KRT19 57.40000, KRT19 577.13950, KRT19 58.18000, KRT19
## 173.69500, KRT19 180.82220, KRT19 232.75320, KRT19 293.32220, KRT19
773.91250,
## KRT19 992.90020, KRT19KRT19, LOC202134 90.27716, LOC202134 116.28997,
LOC202134
## 80.93756, LOC202134 81.95797, LOC202134 95.92870, LOC202134 97.86973,
LOC202134
## 99.14780, LOC202134 103.06420, LOC202134 103.91630, LOC202134 109.43154,
```

```
## LOC202134 153.49023, LOC202134LOC202134, LOC23117 58.95793, LOC23117
63.41986,
## LOC23117 56.92304, LOC23117 57.00729, LOC23117 57.14000, LOC23117
57.58250,
## LOC23117 58.72856, LOC23117 59.18544, LOC23117 59.26586, LOC23117
59.70365,
## LOC23117 60.51843, LOC23117LOC23117, LOC339047 54.32226, LOC339047
55.18711,
## LOC339047 53.41786, LOC339047 54.22306, LOC339047 54.90714, LOC339047
54.96286,
## LOC339047 54.99321, LOC339047 55.83333, LOC339047 56.07547, LOC339047
57.69556,
## LOC339047 57.71951, LOC339047LOC339047, LOC653086 754.31924, LOC653086
## 841.71641, LOC653086 390.88886, LOC653086 735.69129, LOC653086 764.01074,
## LOC653086 870.00770, LOC653086 897.15661, LOC653086 922.11347, LOC653086
## 925.40631, LOC6530861066.72823, LOC6530861203.33167, LOC653086LOC653086,
## MMP11 75.97500, MMP11 136.19640, MMP11 199.35870, MMP11 443.38950, MMP11
## 5787.43480, MMP11 669.80360, MMP111875.99000, MMP112082.50560,
MMP114141.86420,
## MMP118858.71940, MMP11MMP11, PENK 158.16150, PENK 248.20700, PENK
48.20000,
## PENK 54.20000, PENK 55.70000, PENK 60.38330, PENK 61.60000, PENK 89.07890,
PENK
## 255.79600, PENK 699.56800, PENK2788.93790, PENKPENK, PLEC1 117.83730,
PLEC1
## 187.59291, PLEC1 66.99375, PLEC1 68.78959, PLEC1 70.30182, PLEC1 76.05800,
PLEC1
## 79.68368, PLEC1 94.55256, PLEC1 94.90675, PLEC1 104.29716, PLEC1
137.53784,
## PLEC1PLEC1

plot(knnMod)
```



The accuracy seems to be better between 8 and 9 neighbors for classification from what the above plot is displaying.

```
rpartMod <- train(Type ~ ., method='rpart', tuneLength=7, data=trainingSet)

glmMod <- train(Type ~ .,
                method='glm', data=trainingSet)

predKNN <- predict(knnMod, testingSet)
predRPART <- predict(rpartMod, testingSet)
predGLM <- predict(glmMod, testingSet)

length=length(testingSet$Type)

sumKNN <- sum(predKNN==testingSet$Type)
sumRPart <- sum(predRPART==testingSet$Type)
sumGLM <- sum(predGLM==testingSet$Type)

accuracy_KNN <- sumKNN/length
accuracy_RPART <- sumRPart/length
accuracy_GLM <- sumGLM/length

predDF2 <- data.frame(predRF,predKNN,predRPART,predGLM,
                      TYPE=testingSet$Type)
colnames(predDF2) <- c('RandomForest','KNN','Rpart','GLM','TrueValue')

results <- c(round(accuracy_rfMod,2),
```

```
              round(accuracy_KNN,2),
              round(accuracy_RPART,2),
              round(accuracy_GLM,2),
              round(100,2))

results <- as.factor(results)
results <- t(data.frame(results))
colnames(results) <- c('RandomForest','KNN','Rpart','GLM','TrueValue')
Results <- rbind(predDF2, results)
Results
```

```
##         RandomForest  KNN Rpart   GLM TrueValue
## 1                 UL   UL    UL nonUL        UL
## 2                 UL   UL    UL nonUL        UL
## 3                 UL   UL    UL nonUL        UL
## 4                 UL   UL    UL nonUL        UL
## 5                 UL   UL    UL nonUL        UL
## 6                 UL   UL    UL    UL        UL
## 7                 UL   UL    UL nonUL     nonUL
## 8                 UL   UL    UL nonUL     nonUL
## 9                 UL   UL    UL nonUL     nonUL
## 10                UL   UL    UL nonUL     nonUL
## 11                UL   UL    UL nonUL     nonUL
## results         0.55 0.55  0.55  0.55       100
```

I find it difficult to see that there are no changes in values, there are only two classes to predict on 38 samples with 6-20 features, yet every data set and algorithm is producing the same result of 55%. I will see if my other scripts with these algorithms are producing the same results and see if there is a bug in the package libraries or it could be that the mean values in this set were taken from sequence expression values. But I have never seen this type of result using R ever.