Location of MPRA variants in repeats, Vijay's data

Grace Hansen

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Location of MPRA variants in repeats

Variant type	Number of variants	Proportion in repeats	Proportion in Alus
EMVar	68	0.4853	0.2059
Enhancer	339	0.5015	0.2301
Nonsignificant	2223	0.5888	0.2519

Retrovirus types in MPRA

Types of repeats in EMVars:

RV	count
SINE	15
LINE	9
LTR	8
Simple_repeat	1
DNA	0
DNA?	0
snRNA	0
Retroposon	0

Sub-types of repeats in EMVars:

RV subtype	count
AluY	4
AluSp	2
AluYk3	2
AluJo	1
AluSg	1
AluSx	1
AluYf1	1
AluYk2	1
AluYm1	1
$FLAM_C$	0
HAL1	0
HERV9	0
$L1M5_orf2$	0
$L1MA4_3end$	0

RV subtype	count
L1MC1_3end	0
$L1MC4_3end$	0
$L1ME1_3end$	0
$L1ME4b_3end$	0
$L1MEd_5end$	0
L1P1_orf2	0
$L1PA4_3end$	0
LTR12C	0
LTR12E	0
LTR53	0
MLT1E2	0
THE1B	0
THE1-int	0

Plot results with chi-square p values

Are there more repeats and Alus in repeats than expected by chance?

```
## [1] "Repeat Chi square observed vs expected tables:"
```

```
##
             repeats no repeats
## EMVars
                  33
                 170
## enhancers
                             169
## nonsig
                1309
                             914
##
                repeats no repeats
## EMVars
                           28.90646
               39.09354
## enhancers
             194.89278
                         144.10722
                         944.98631
             1278.01369
## nonsig
## [1] "Alu Chi square observed vs expected tables:"
##
             repeats no repeats
## EMVars
                  14
                              54
## enhancers
                  78
                             261
                 560
## nonsig
                            1663
##
               repeats no repeats
## EMVars
              16.85779
                         51.14221
## enhancers 84.04106 254.95894
## nonsig
             551.10114 1671.89886
## pdf
##
     2
## pdf
##
     2
```

Expression of Alu elements

Are sequences containing Alu elements more highly expressed than sequences not containing Alu elements?

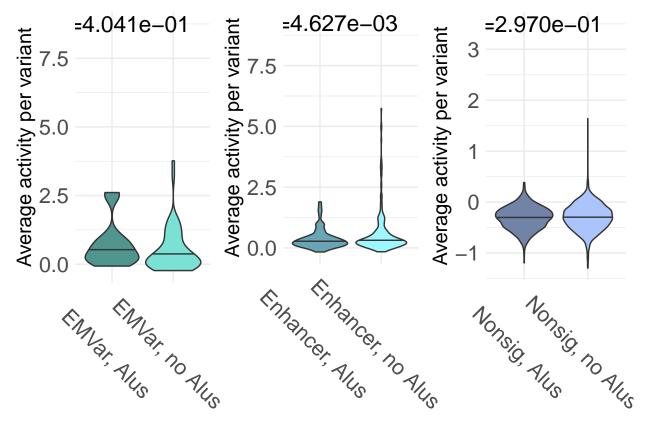
[1] "qnorm from nonsignificant sequences without Alu elements"

```
Min. 1st Qu. Median
                             Mean 3rd Qu.
## -1.2955 -0.4690 -0.2969 -0.2991 -0.1289
                                          1.6376
## [1] "qnorm from nonsignificant sequences with Alu elements"
     Min. 1st Qu. Median
                             Mean 3rd Qu.
## -1.1930 -0.4693 -0.2916 -0.3110 -0.1582 0.3839
## [1] "qnorm from enhancer sequences without Alu elements"
     Min. 1st Qu. Median
##
                             Mean 3rd Qu.
                                             Max.
## -0.1584 0.1520 0.2983 0.5848 0.6244 5.7360
## [1] "qnorm from enhancer sequences with Alu elements"
     Min. 1st Qu. Median
                             Mean 3rd Qu.
## -0.1665 0.1147 0.2770 0.3801 0.4592 1.8944
## [1] "qnorm from EMVar sequences without Alu elements"
            1st Qu.
                      Median
##
      Min.
                                 Mean 3rd Qu.
                     0.19763
                              0.51134
## -0.23262
            0.01322
                                       0.87895
                                                3.76455
## [1] "qnorm from EMVar sequences with Alu elements"
##
      Min. 1st Qu.
                      Median
                                 Mean 3rd Qu.
                                                   Max.
## -0.06914 0.17000 0.48447 0.72253 0.82545 2.61034
```

Significance and visualization

In the plots below, you can see that the Alu-containing sequences have higher expression in enhancers and EMVars, but this isn't true for nonsignificant sequences.

```
## Warning in regularize.values(x, y, ties, missing(ties)): collapsing to unique
## 'x' values
```



 $\hbox{\tt \#\# Warning in regularize.values(x, y, ties, missing(ties)): collapsing to unique}$

'x' values

pdf

2