## Class 10: Genome Informatics

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2/17/2022

## Examine 1000 Genome Data

Q5: What proportion of the Mexican Ancestry in Los Angeles sample population (MXL) are homozygous for the asthma associated SNP (G|G)?

14.06

```
# Read genotype file from Ensemble
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
round(table(mxl$Genotype..forward.strand.)/nrow(mxl)*100,2)

##
## A|A A|G G|A G|G
## 34.38 32.81 18.75 14.06</pre>
```

What about a different population? Here we take the British in England and Scotland (GBR)

```
gbr <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
round(table(gbr$Genotype..forward.strand.)/nrow(gbr) * 100,2)</pre>
```

```
##
## A|A A|G G|A G|G
## 25.27 18.68 26.37 29.67
```

## Expression by Genotype analysis

I want read my RNA-Seq expression results into R. This file is not a CSV but rather has fields separated by space.

```
x <- read.table("rs8067378_ENSG00000172057.6.txt")
x</pre>
```

```
## sample geno exp

## 1 HG00367 A/G 28.96038

## 2 NA20768 A/G 20.24449

## 3 HG00361 A/A 31.32628

## 4 HG00135 A/A 34.11169

## 5 NA18870 G/G 18.25141
```

```
## 6
       NA11993
                A/A 32.89721
## 7
       HG00256
                 A/G 31.48736
       NA18498
## 8
                 A/A 47.64556
## 9
       HG00327
                 G/G 17.67473
## 10
       HG00115
                 A/G 33.85374
       NA20806
## 11
                 A/G 16.29854
                 A/G 19.73450
       HG00278
## 12
## 13
       NA20585
                 A/A 30.71355
## 14
       NA19137
                 A/G 13.96175
## 15
       HG00235
                 A/A 25.44983
## 16
       NA20798
                 A/A 34.24915
       NA12546
                 G/G 18.55622
## 17
## 18
       NA19116
                 A/A 35.15014
       HG00381
                 A/G 18.40351
## 19
## 20
       NA18488
                 G/G 23.10383
## 21
       HG00259
                 A/G 34.21985
## 22
                 A/G 23.32404
       HG00177
## 23
       NA19214
                 G/G 30.94554
## 24
                 A/A 24.54684
       NA19247
## 25
       NA19098
                 A/G 23.18606
## 26
       NA20589
                 A/G 18.15997
## 27
       NA19207
                 A/A 49.39612
       HG00112
                 G/G 21.14387
## 28
                 G/G 18.39547
       NA20518
## 29
## 30
       HG00335
                 A/A 28.20755
## 31
       NA19119
                 G/G 12.02809
## 32
       HG00247
                 G/G 17.44761
##
   33
       NA12155
                 A/G 28.03580
##
   34
       NA20771
                 A/G 30.65270
##
  35
       NA20758
                 G/G 29.82254
## 36
       HG00121
                 A/G 20.51327
## 37
       NA20759
                 A/A 28.56199
##
  38
       NA20816
                 A/G 29.72309
       NA20542
                 A/G 22.50789
## 39
## 40
       NA18511
                 A/G 31.68959
## 41
                 G/G 23.01983
       NA12249
## 42
       NA11830
                 A/G 28.76435
## 43
       NA19159
                 A/G 35.85543
## 44
       NA20778
                 A/G 37.62403
       NA18908
                 A/G 20.54885
## 45
                 G/G 13.42470
       HG00320
## 46
## 47
       NA11843
                 G/G 22.65437
## 48
       HG00105
                 A/A 51.51787
## 49
       NA20588
                 G/G 11.07445
                 G/G 28.35841
## 50
       NA20510
## 51
       NA12342
                 A/G 31.04941
## 52
       HG00249
                 A/G 18.94583
## 53
       NA11894
                 A/A 38.10956
## 54
       HG00240
                 A/G 32.29483
## 55
       HG00132
                 A/A 31.13741
## 56
                 G/G 28.79371
       HG00118
## 57
       NA18520
                 G/G 27.08956
## 58
       NA18508
                 A/G 27.81775
## 59
       HG00353 A/G 19.89903
```

```
## 60
      NA20792 A/G 48.03410
## 61
       NA12234
                G/G 16.11138
                A/A 39.12999
       HG00377
## 63
       NA19143
                A/G 27.90313
## 64
       NA20787
                A/G 36.47949
                A/G 20.03116
## 65
       NA20513
       HG00243
                A/G 29.65063
## 66
## 67
       NA19172
                A/A 32.44173
## 68
       NA06994
                A/G 34.92257
## 69
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                A/G 16.71385
## 70
       HG00337
                A/G 16.68151
       NA20503
                A/G 25.71008
## 71
## 72
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                G/G 30.18323
## 73
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## 74
       NA19235
                A/G 11.60808
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       HG00382
                A/G 19.30953
## 76
       NA20544
                A/A 34.03260
## 77
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       HG00313
                A/G 20.49040
## 78
## 79
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                G/G 19.52301
## 80
       NA20754
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                A/G 15.20045
                A/A 36.27151
## 82
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       NA06986
                A/G 20.07459
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## 84
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## 85
       NA12058
                G/G 26.56808
## 86
       NA20507
                 A/G 19.10884
## 87
       NA12777
                A/G 24.81087
## 88
       NA12144
                A/G 33.22193
## 89
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                G/G 17.34076
## 90
       HG00123
                A/G 33.40835
## 91
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                A/G 22.38996
## 92
       HG00183
                G/G 10.74263
## 93
       HG00109
                G/G 16.66051
## 94
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                A/G 31.31626
## 95
                A/G 9.36055
       NA12273
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                A/A 26.10355
## 97
       HG00324
                A/A 19.48106
## 98
       HG00365
                A/G 23.17937
## 99
       NA20520
                 A/A 38.77623
## 100 NA19189
                 A/G 30.63079
## 101 HG00155
                A/G 19.10420
## 102 HG00111
                A/A 40.82922
## 103 NA12827
                 A/G 25.70962
## 104 NA18517
                G/G 29.01720
## 105 NA20801
                G/G 20.69333
## 106 NA20529
                G/G 21.15677
## 107 NA18909
                A/G 38.34531
## 108 HG00173
                A/G 19.03976
## 109 HG00349
                G/G 18.58691
## 110 HG00234
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## 111 NA19248
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## 112 NA20810
                A/A 46.50527
## 113 HG00255 A/G 28.81770
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## 115 NA20537
               G/G 21.12823
## 116 NA18912
                A/G 42.75662
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## 118 HG00152
                G/G 19.37093
## 119 NA20783
                G/G 31.42162
## 120 NA12154
                A/G 25.61662
## 121 HG00236
                A/A 33.07320
## 122 NA19146
                A/A 25.47283
## 123 HG00312
                A/G 26.48467
## 124 HG00148
               A/G 28.02486
## 125 HG00364
                A/G 24.23377
## 126 HG00311
                A/G 21.03717
## 127 NA11881
                A/A 29.50655
## 128 HG00185 G/G 16.67764
## 129 NA20807
                A/G 33.51752
## 130 NA19184
                A/G 20.73493
## 131 HG00133
               A/G 33.55650
## 132 NA20531
                G/G 19.08659
## 133 NA19138
                A/A 27.48438
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                A/G 36.62034
## 135 HG00277
                G/G 21.55001
## 136 NA18858
                A/G 40.06318
## 137 HG00375
                A/G 33.92744
## 138 HG00127
                A/G 21.02084
## 139 NA19099
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## 140 HG00336
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## 145 NA20797
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## 146 NA12872
                A/G 30.03549
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## 162 NA12749
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## 163 NA19190
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## 164 NA06985
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## 165 HG00178
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## 166 NA10851 G/G 23.53572
## 167 HG00371 A/A 19.14544
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## 170 HG00116
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## 172 NA19096
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## 173 NA20800
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## 174 HG00102
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## 175 NA19236
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## 177 NA20521
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## 178 HG00345
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## 181 NA12830
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## 182 HG00359
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## 192 HG00180
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## 194 HG00343
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## 195 HG00139
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## 197 HG00321
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## 199 HG00232
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## 204 HG00151
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## 207 NA19149
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## 213 NA11832 A/G 34.47373
## 214 HG00323
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## 215 NA18916
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## 217 HG00100
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## 221 NA20532 A/G 21.76610
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## 226 NA20790
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## 227 NA20512
                A/A 37.94544
                A/A 29.15536
## 228 HG00268
## 229 HG00380
                A/A 28.85309
## 230 NA12761
                A/A 38.57101
## 231 HG00384
                A/G 29.49417
## 232 NA20796
                G/G 23.92355
## 233 NA12399
                G/G 9.55902
## 234 HG00310
                A/G 29.55520
## 235 HG00096
                A/A 30.89365
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## 237 NA20752
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## 238 NA19107
                A/G 30.40382
## 239 HG00099
                G/G 12.35836
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                A/A 39.31537
## 241 NA19114
                G/G 22.53910
## 242 HG00376
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## 243 NA19092
                A/A 35.26739
## 244 HG00130
                A/G 28.50982
## 245 HG00158
                A/A 22.37043
## 246 HG00269
                A/A 28.46943
## 247 NA19210
                G/G 21.98118
## 248 HG00258
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## 249 NA19256
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                A/G 31.10134
                A/G 35.99067
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## 255 NA11920
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## 256 HG00326
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## 257 NA12347
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## 258 NA12716
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## 259 HG00142
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## 260 HG00309
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## 261 HG00315
                G/G 26.56993
## 262 HG00338
                A/G 23.79292
## 263 NA11995
                A/A 32.59723
                A/A 36.02549
## 264 NA19209
## 265 NA20540
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## 266 NA12890
                A/A 28.38114
## 267 HG00250
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## 268 NA20769
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## 269 HG00138
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## 270 NA19200
                A/A 51.30170
## 271 NA19144
                G/G 24.85165
## 272 NA12815
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## 274 HG00350 A/G 29.54042
## 275 NA12383 A/A 28.14811
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## 278 NA06984
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## 280 NA19175
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## 282 NA12044
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## 283 NA18519
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## 284 NA20799
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## 285 NA20535
                G/G 22.53720
## 286 NA19141
                A/G 28.72738
## 287 HG00260
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## 288 HG00372
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## 290 NA07357
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## 291 NA20543
                A/G 34.14567
## 292 HG00261
                G/G 20.07363
## 293 HG00273
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## 294 NA12341
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## 295 HG00245
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## 297 NA20757
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## 298 NA11930
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## 299 HG00358
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## 302 NA20773
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## 306 HG00117
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## 307 NA19121
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## 308 NA20515
                G/G 18.07151
## 309 HG00355
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## 310 NA12775
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                A/G 16.12745
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## 314 NA10847
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## 315 NA19102
                A/G 13.08172
## 316 NA12400
                G/G 22.14277
## 317 NA18487
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## 318 NA19093
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## 322 HG00160
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## 323 NA20766
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## 324 NA12717
                A/G 7.07505
## 325 HG00125
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## 326 HG00171
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## 329 NA20826 A/G 18.24345
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## 331 HG00272
## 332 NA12340
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## 336 NA12842
## 337 HG00146
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## 342 HG00114
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## 359 HG00157
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## 360 HG00262
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## 364 NA18910
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## 365 NA20795
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## 373 NA12275
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## 374 NA20514
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## 375 HG00351
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## 376 HG00186
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## 377 NA20586
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## 378 HG00275
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## 379 HG00325
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                G/G 24.80823
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## 383 HG02215 G/G 18.28089
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## 388 NA12778
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## 389 NA18861
## 390 NA20539
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## 391 NA11931
## 392 NA20812
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## 393 HG00120
                G/G 21.09502
## 394 HG00103
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## 395 HG00328
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## 396 NA20774
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## 397 NA18873
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## 399 HG00143
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## 400 HG00145
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## 401 NA19225
                A/A 26.56050
## 402 NA12829
                A/G 28.98200
## 403 HG00137
                A/G 34.31875
## 404 NA20524
                A/G 26.40231
## 405 HG00379
                A/A 21.87746
## 406 NA18505
                A/G 21.67621
## 407 HG01334
                A/G 27.56805
## 408 NA18907
                A/A 33.42582
## 409 NA19204
                A/A 25.38406
## 410 NA12874
                A/G 16.16277
## 411 NA20506
                A/G 18.28963
## 412 NA20770
                A/A 18.20442
## 413 NA12776
               A/G 30.55183
## 414 NA18934
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## 415 NA19153
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## 416 HG00356
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## 417 NA12283
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## 418 HG00284
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## 419 NA12489
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## 420 HG00104
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## 421 NA20582
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## 422 NA11840
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## 423 HG00383
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## 425 NA20802
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## 426 NA20756
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## 427 NA19113
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## 428 NA12889
## 429 NA12718
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## 430 HG00266
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## 437 NA19185 A/G 28.93651
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## 440 NA19257
## 441 NA12413
               A/G 39.43243
## 442 HG00159
               A/A 23.99631
## 443 NA20811
               A/A 11.39643
## 444 HG00149
              A/G 23.91465
## 445 NA19223
               A/G 20.97560
## 446 NA07346
               G/G 16.56929
## 447 NA20536
               A/G 20.02507
## 448 HG01791
               A/A 35.24632
## 449 HG00271
               A/G 33.44170
## 450 HG00373 A/G 17.32813
## 451 HG00182 A/A 23.38376
## 452 HG00110 A/G 32.61856
## 453 NA20819 A/G 36.77906
## 454 HG00154
               G/G 16.69044
## 455 HG00330
               A/G 16.84776
## 456 NA12750
               A/A 34.94395
## 457 HG00233
               G/G 25.08880
## 458 HG00131
               G/G 32.78519
## 459 HG00108
               A/A 31.92036
               A/G 31.53069
## 460 HG00119
## 461 NA19130
               A/A 44.27738
## 462 HG00239
              A/G 23.18250
```

First try at this question. Is the mean expression different based on genotype?

```
x\$geno == "G/G"
```

```
[1] FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE
##
   [13] FALSE FALSE FALSE
                              TRUE FALSE FALSE TRUE FALSE FALSE
                                                               TRUE FALSE
##
   [25] FALSE FALSE FALSE TRUE
                              TRUE FALSE TRUE TRUE FALSE FALSE
                                                               TRUE FALSE
##
   [37] FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE TRUE TRUE FALSE
        TRUE TRUE FALSE FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE
        TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE TRUE
   [61]
##
   [73]
        TRUE FALSE FALSE FALSE
                              TRUE FALSE TRUE FALSE FALSE FALSE FALSE
##
        TRUE FALSE FALSE FALSE
                              TRUE FALSE FALSE
   [85]
                                               TRUE
                                                    TRUE FALSE FALSE FALSE
   [97] FALSE FALSE FALSE FALSE FALSE FALSE
                                               TRUE
                                                    TRUE
                                                         TRUE FALSE FALSE
## [109] TRUE TRUE
                  TRUE FALSE FALSE TRUE
                                         TRUE FALSE
                                                    TRUE
                                                         TRUE
                                                               TRUE FALSE
  [121] FALSE FALSE FALSE FALSE FALSE FALSE
                                               TRUE FALSE FALSE FALSE
## [133] FALSE FALSE TRUE FALSE FALSE FALSE
                                              TRUE FALSE FALSE TRUE FALSE
## [145] FALSE FALSE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE
## [157] FALSE FALSE
                  TRUE FALSE FALSE FALSE
                                        TRUE FALSE FALSE
                                                         TRUE FALSE FALSE
## [169] FALSE TRUE TRUE
                        TRUE FALSE FALSE TRUE FALSE FALSE
                                                          TRUE FALSE FALSE
## [181] FALSE FALSE FALSE FALSE FALSE FALSE FALSE
                                                         TRUE FALSE FALSE
## [193] TRUE
             TRUE
                   TRUE FALSE FALSE FALSE
                                         TRUE FALSE
                                                    TRUE FALSE FALSE FALSE
                   TRUE FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE
## [205] FALSE FALSE
## [217] FALSE TRUE FALSE FALSE FALSE FALSE TRUE
                                                   TRUE FALSE FALSE FALSE
## [229] FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE TRUE FALSE
        TRUE FALSE FALSE FALSE FALSE TRUE FALSE TRUE FALSE TRUE FALSE
## [241]
## [253] TRUE TRUE FALSE FALSE FALSE
                                         TRUE FALSE TRUE FALSE FALSE
## [265] FALSE FALSE TRUE TRUE FALSE FALSE TRUE TRUE FALSE FALSE FALSE
```

```
## [277] FALSE FALSE TRUE FALSE FALSE TRUE FALSE TRUE FALSE TRUE TRUE
## [289] FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE TRUE FALSE
## [301] FALSE FALSE FALSE FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE
## [313] FALSE TRUE FALSE TRUE FALSE FALSE
## [325] FALSE FALSE FALSE FALSE
                                                                                 TRUE FALSE FALSE FALSE FALSE FALSE
## [337] FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE
## [349] FALSE FALSE TRUE FALSE FALSE TRUE
                                                                                                        TRUE
                                                                                                                       TRUE FALSE FALSE FALSE
## [361] TRUE TRUE FALSE
                                                       TRUE FALSE FALSE FALSE
                                                                                                                       TRUE FALSE FALSE FALSE
                                                       TRUE FALSE TRUE
## [373] TRUE FALSE TRUE
                                                                                            TRUE
                                                                                                        TRUE
                                                                                                                       TRUE FALSE
                                                                                                                                              TRUE FALSE
## [385] TRUE FALSE FALSE FALSE FALSE
                                                                                            TRUE FALSE
                                                                                                                      TRUE FALSE FALSE FALSE
## [397] FALSE FALSE
## [409] FALSE FALSE
## [421] TRUE FALSE FALSE FALSE FALSE FALSE TRUE FALSE FALSE FALSE FALSE
## [433] FALSE FALSE TRUE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE
## [445] FALSE TRUE FALSE FALSE FALSE FALSE FALSE FALSE FALSE FALSE
## [457] TRUE
                              TRUE FALSE FALSE FALSE
summary(x[x$geno =="G/G",]$exp)
##
            Min. 1st Qu. Median
                                                             Mean 3rd Qu.
                                                                                              Max.
##
          6.675 16.903 20.074
                                                        20.594
                                                                         24.457 33.956
Now we will look at the other genotypes
table(x$geno)
##
## A/A A/G G/G
## 108 233 121
summary(x[x\$geno == "A/A",3])
##
            Min. 1st Qu. Median
                                                             Mean 3rd Qu.
                                                                                              Max.
##
          11.40
                          27.02
                                          31.25
                                                           31.82
                                                                            35.92
                                                                                            51.52
summary(x[x\$geno == "A/G",3])
##
            Min. 1st Qu.
                                        Median
                                                             Mean 3rd Qu.
                                                                                              Max.
          7.075 20.626
                                        25.065 25.397 30.552
summary(x[x\$geno == "G/G",3])
##
            Min. 1st Qu.
                                        Median
                                                             Mean 3rd Qu.
                                                                                              Max.
##
          6.675 16.903 20.074 20.594
                                                                       24.457
```

## Make a summary overview figure

Make a boxplot figure...

```
library(ggplot2)
ggplot(x) + aes(geno, exp, fill = geno) + geom_boxplot(notch = TRUE)
```

