## Class 12 HW

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# Reading the final file into R and interpreting $\mathbf{Q}\mathbf{1}\mathbf{3}$

```
url<-"https://bioboot.github.io/bggn213_W19/class-material/rs8067378_ENSG00000172057.6.txt"
data<-read.table(url)
data</pre>
```

```
sample geno
                     exp
   HG00367 A/G 28.96038
1
  NA20768 A/G 20.24449
2
3
   HG00361 A/A 31.32628
   HG00135 A/A 34.11169
5
   NA18870 G/G 18.25141
   NA11993 A/A 32.89721
7
   HG00256 A/G 31.48736
   NA18498 A/A 47.64556
8
9
   HG00327 G/G 17.67473
10 HG00115 A/G 33.85374
11 NA20806 A/G 16.29854
12 HG00278 A/G 19.73450
13 NA20585 A/A 30.71355
14 NA19137 A/G 13.96175
15 HG00235 A/A 25.44983
16 NA20798 A/A 34.24915
17 NA12546 G/G 18.55622
18 NA19116 A/A 35.15014
19 HG00381 A/G 18.40351
20 NA18488 G/G 23.10383
21 HG00259 A/G 34.21985
22 HG00177 A/G 23.32404
23 NA19214 G/G 30.94554
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24 NA19247 A/A 24.54684 NA19098 A/G 23.18606 25 26 NA20589 A/G 18.15997 27 NA19207 A/A 49.39612 G/G 21.14387 28 HG00112 NA20518 G/G 18.39547 29 30 HG00335 A/A 28.20755 31 NA19119 G/G 12.02809 HG00247 G/G 17.44761 32 33 NA12155 A/G 28.03580 34 NA20771 A/G 30.65270 NA20758 G/G 29.82254 35 36 HG00121 A/G 20.51327 37 NA20759 A/A 28.56199 38 NA20816 A/G 29.72309 39 NA20542 A/G 22.50789 40 NA18511 A/G 31.68959 41 NA12249 G/G 23.01983 42 NA11830 A/G 28.76435 43 NA19159 A/G 35.85543 44 NA20778 A/G 37.62403 45 NA18908 A/G 20.54885 46 HG00320 G/G 13.42470 G/G 22.65437 47 NA11843 48 HG00105 A/A 51.51787 G/G 11.07445 49 NA20588 NA20510 G/G 28.35841 50 51 NA12342 A/G 31.04941 HG00249 52 A/G 18.94583 53 NA11894 A/A 38.10956 54 HG00240 A/G 32.29483 55 HG00132 A/A 31.13741 56 HG00118 G/G 28.79371 57 NA18520 G/G 27.08956 58 NA18508 A/G 27.81775 59 HG00353 A/G 19.89903 60 NA20792 A/G 48.03410 NA12234 G/G 16.11138 61 A/A 39.12999 62 HG00377 63 NA19143 A/G 27.90313 NA20787 A/G 36.47949 64 NA20513 A/G 20.03116 65

HG00243 A/G 29.65063

66

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68
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   NA18510
             A/G 16.71385
69
70
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            A/G 25.71008
71
   NA20503
             G/G 26.61928
72
   NA19152
73
   NA20761
             G/G 30.18323
74
   NA19235
             A/G 11.60808
   HG00382
            A/G 19.30953
75
76
   NA20544
             A/A 34.03260
77
             G/G 19.40790
   NA18923
             A/G 20.49040
78
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79
   HG00238
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80
   NA20754
             A/G 22.37224
81
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   NA18868
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83
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   NA20507
             A/G 19.10884
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87
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             A/G 33.22193
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89
   HG00129
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93
   HG00109
94
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             A/G 9.36055
95
   NA12273
96
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97
   HG00324
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98
   HG00365
             A/G 23.17937
99
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             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
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115 NA20537 G/G 21.12823
116 NA18912 A/G 42.75662
117 HG00332 G/G 18.61268
118 HG00152 G/G 19.37093
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120 NA12154 A/G 25.61662
121 HG00236 A/A 33.07320
122 NA19146 A/A 25.47283
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124 HG00148 A/G 28.02486
125 HG00364 A/G 24.23377
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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
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134 NA19206 A/G 36.62034
            G/G 21.55001
135 HG00277
136 NA18858 A/G 40.06318
137 HG00375
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138 HG00127 A/G 21.02084
139 NA19099 A/G 29.95687
140 HG00336 G/G 8.29591
           A/G 25.80393
141 HG00097
142 HG00267
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143 NA20581 G/G 12.58869
144 NA12286
           A/G 34.79575
145 NA20797 A/G 34.57705
146 NA12872 A/G 30.03549
147 HG00360
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148 NA20530 A/G 27.22300
149 NA12348 A/G 24.35621
150 NA20538 G/G 17.34109
151 NA12760 A/G 22.86793
152 NA12763 A/G 23.19511
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157 NA11829 A/G 33.74015
158 NA11992 A/G 24.08401
159 HG00141 G/G 25.55413
160 NA19150 A/G 26.39419
161 NA20828 A/G 32.33359
162 NA12749
           A/A 28.91526
163 NA19190 G/G 24.45672
164 NA06985
           A/G 11.36287
165 HG00178
           A/G 21.16515
166 NA10851
           G/G 23.53572
167 HG00371
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168 NA20541 A/G 17.21277
169 NA12004
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170 HG00116 G/G 22.48273
171 NA12272 G/G 14.66862
172 NA19096 G/G 33.95602
173 NA20800
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174 HG00102 A/A 31.17067
175 NA19236 G/G 18.26466
176 HG00264
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177 NA20521 A/A 27.87464
178 HG00345
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           A/G 11.97590
181 NA12830
182 HG00359
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183 NA07051
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184 NA20516 A/G 33.32411
185 HG00128
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186 NA20534
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187 NA11892
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188 NA20804 A/A 36.51922
189 NA11994 A/G 30.83577
190 HG00156
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191 NA12843 A/G 23.63709
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193 HG00282 G/G 19.14766
194 HG00343 G/G 12.57599
195 HG00139 G/G 22.28749
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203 NA07056 A/G 15.92557
204 HG00151 A/G 32.54150
205 NA19129
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206 NA20517 A/G 22.40203
207 NA19149 G/G 16.07627
208 HG00341 A/G 27.41638
209 HG00274 A/G 31.99645
210 HG00106 A/G 30.05415
211 HG00189 G/G 14.80495
212 HG00252 A/G 20.01602
213 NA11832 A/G 34.47373
214 HG00323 A/A 22.44576
215 NA18916 A/A 37.06379
           A/G 28.75978
216 NA18867
217 HG00100 A/A 35.67637
218 HG00126 G/G 23.46573
219 NA20813
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220 NA20504 A/G 15.71646
221 NA20532 A/G 21.76610
222 NA12812 A/G 9.62656
223 HG00244 A/G 28.53965
224 HG00265 G/G 28.97074
225 HG00378 G/G 27.78837
226 NA20790 A/A 50.16704
227 NA20512 A/A 37.94544
228 HG00268 A/A 29.15536
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
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234 HG00310 A/G 29.55520
235 HG00096 A/A 30.89365
236 NA19147 A/G 19.44178
237 NA20752 A/G 21.43751
238 NA19107 A/G 30.40382
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241 NA19114 G/G 22.53910
242 HG00376 A/A 31.43743
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 A/A 30.15636
249 NA19256 A/G 21.48847
250 HG00276 G/G 16.40569
251 HG00331 A/G 31.10134
252 NA12751 A/G 35.99067
253 HG00181 G/G 25.21931
254 HG00346 G/G 24.32857
255 NA11920
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256 HG00326 A/G 26.28329
257 NA12347 A/A 35.88457
258 NA12716 A/G 20.72639
259 HG00142 G/G 19.42882
260 HG00309 A/G 21.09140
261 HG00315 G/G 26.56993
262 HG00338 A/G 23.79292
263 NA11995 A/A 32.59723
264 NA19209 A/A 36.02549
265 NA20540 A/A 23.86454
266 NA12890 A/A 28.38114
267 HG00250 G/G 13.34557
268 NA20769 G/G 16.60507
269 HG00138
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270 NA19200 A/A 51.30170
271 NA19144 G/G 24.85165
272 NA12815 G/G 21.56943
273 NA12043 A/G 18.79569
274 HG00350 A/G 29.54042
275 NA12383 A/A 28.14811
276 NA19201 A/G 18.78700
277 HG00187 A/G 21.41071
278 NA06984 A/A 29.18390
279 NA20508 A/G 21.29782
280 NA19175 G/G 23.95528
281 NA20815 A/G 33.91853
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283 NA18519 G/G 16.18962
284 NA20799 A/G 17.14895
285 NA20535 G/G 22.53720
286 NA19141 A/G 28.72738
287 HG00260 G/G 26.04123
288 HG00372 G/G 6.67482
289 NA07347 A/G 37.73840
290 NA07357 A/A 27.09760
291 NA20543 A/G 34.14567
292 HG00261 G/G 20.07363
293 HG00273 G/G 19.76527
294 NA12341 A/G 15.36874
295 HG00245 A/G 29.50350
296 NA19198 A/G 25.70400
297 NA20757 A/G 20.07219
298 NA11930 A/A 33.89656
299 HG00358 G/G 18.50772
300 NA18933 A/G 24.53928
301 HG00242 A/G 17.84487
302 NA20773 A/G 23.35766
303 NA12282 A/G 15.71243
304 NA19131 A/A 33.48253
305 NA18499 A/A 15.43178
306 HG00117 A/A 29.45277
307 NA19121 G/G 20.14146
308 NA20515 G/G 18.07151
309 HG00355 A/G 19.89034
310 NA12775 A/G 25.37234
311 NA12005 A/G 16.12745
312 NA11893 A/G 24.18529
313 NA20808 A/G 21.97051
314 NA10847 G/G 6.94390
315 NA19102 A/G 13.08172
316 NA12400 G/G 22.14277
317 NA18487 A/G 32.00764
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319 HG00342 G/G 14.23742
320 NA19160 A/G 29.74443
321 NA19095 A/G 27.88354
322 HG00160 A/A 26.80283
323 NA20766 A/G 11.12451
324 NA12717 A/G 7.07505
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325 HG00125 A/G 23.13726
326 HG00171 A/G 21.09331
327 NA12873 A/G 8.20002
328 NA20525 A/G 20.62572
329 NA20826 A/G 18.24345
330 HG00136 G/G 19.85388
331 HG00272 A/G 11.13478
332 NA12340 A/A 43.51943
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334 HG00369 A/G 22.24289
335 NA20803 A/G 24.67325
336 NA12842 A/G 41.03924
337 HG00146 A/A 45.80808
338 HG01790 A/G 33.31795
339 NA20809 A/G 27.98844
340 NA20765 G/G 27.73467
341 HG00362
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342 HG00114 A/G 31.57994
343 NA18917 A/A 24.87330
344 NA18502 G/G 19.02064
345 HG00150 A/G 36.73337
346 NA20527 A/A 29.99549
347 HG00179 A/G 18.45322
348 NA20805 A/A 26.68589
349 NA19117 A/G 23.60431
350 HG00285 A/G 24.33489
351 NA20772 G/G 14.49816
352 NA19213
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353 HG00344 A/G 22.75684
354 NA12156 A/A 39.37193
355 HG00257
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356 NA18486 G/G 20.84709
357 HG00188 G/G 10.77316
358 HG00366
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359 HG00157
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360 HG00262 A/A 41.23635
361 HG00280 G/G 12.82128
362 HG00308 G/G 16.90256
363 NA11831 A/G 25.34866
364 NA18910 G/G 29.60045
365 NA20795 A/G 25.06486
366 HG00231 A/G 36.78028
367 NA19197 A/G 30.67131
```

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368 HG00101 A/A 27.13936
369 HG00281 G/G 14.81945
370 NA20760
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371 HG00176 A/A 28.34688
372 NA18489 A/G 37.82860
373 NA12275 G/G 17.46326
374 NA20514 A/A 15.42908
375 HG00351 G/G 23.26922
376 HG00186 G/G 21.39806
377 NA20586
            A/G 25.44086
378 HG00275 G/G 18.06320
379 HG00325 G/G 15.91528
380 NA19118
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381 HG00124
           G/G 26.04514
382 NA20785
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383 HG02215 G/G 18.28089
384 HG00253
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385 HG00134 G/G 23.24907
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390 NA20539
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391 NA11931
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392 NA20812 A/G 28.69506
393 HG00120
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394 HG00103
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395 HG00328
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397 NA18873 A/G 25.81562
398 NA20502 A/G 22.49429
399 HG00143 A/G 26.88264
400 HG00145
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401 NA19225
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402 NA12829
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403 HG00137 A/G 34.31875
404 NA20524 A/G 26.40231
405 HG00379
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406 NA18505 A/G 21.67621
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408 NA18907 A/A 33.42582
409 NA19204 A/A 25.38406
410 NA12874 A/G 16.16277
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413 NA12776 A/G 30.55183
414 NA18934 A/G 20.70871
415 NA19153 A/G 17.66476
416 HG00356 A/G 22.79543
417 NA12283 A/G 24.03419
418 HG00284 A/G 18.02351
419 NA12489 A/G 21.63102
420 HG00104
           A/A 21.62336
421 NA20582 G/G 24.74366
422 NA11840 A/G 27.54976
423 HG00383 A/G 14.79717
424 NA20786
           A/A 35.80093
425 NA20802 A/G 25.34921
426 NA20756 A/A 32.26844
427 NA19113 A/G 21.34916
428 NA12889 G/G 27.40521
429 NA12718 A/G 21.20080
430 HG00266 A/G 28.36006
431 NA12287
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432 HG00319 A/G 25.56306
433 NA12762 A/A 34.40756
434 HG00334
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435 NA12006 G/G 24.85772
436 NA19108 G/G 23.08482
437 NA19185 A/G 28.93651
438 HG00246
           A/G 31.79897
439 NA12045 A/G 30.80067
440 NA19257
           A/G 33.95134
441 NA12413
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442 HG00159
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443 NA20811
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444 HG00149
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445 NA19223
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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

460 HG00119 A/G 31.53069

461 NA19130 A/A 44.27738

462 HG00239 A/G 23.18250
```

We aim to determine the sample size for each genotype (A|G, A|A, and G|G) and their corresponding median expression levels.

```
# Reading the data summary(data)
```

```
sample
                        geno
                                              exp
                                                : 6.675
Length: 462
                    Length:462
                                        Min.
Class : character
                    Class : character
                                        1st Qu.:20.004
Mode :character
                    Mode
                          :character
                                        Median :25.116
                                                :25.640
                                        Mean
                                        3rd Qu.:30.779
                                        Max.
                                                :51.518
```

Now, individually:

```
table(data$geno)
```

```
A/A A/G G/G
108 233 121
```

A13a. Therefore, the sample size for the A|A genotype is 108, for A/G is 233, and for G|G is 121.

Their corresponding median expression levels can be determined by:

#### summary(data\$exp)

```
Min. 1st Qu. Median Mean 3rd Qu. Max. 6.675 20.004 25.116 25.640 30.779 51.518
```

```
boxplot_stats<- boxplot(exp~geno, data=data, plot=F)
medians<- boxplot_stats$stats[3,]

names(medians) <- levels(data$geno)
medians</pre>
```

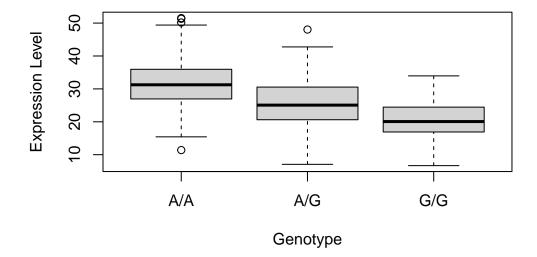
#### [1] 31.24847 25.06486 20.07363

A13b. Their median expression levels are approximately 31.25, 25.06, and 20.07, respectively.

Q14 To make a boxplot, we will use the following code. With this, we aim to make an inference about the two homozygous genotypes.

```
boxplot(exp~geno, data=data,
main = "Gene Expression by Genotype",
xlab = "Genotype",
ylab = "Expression Level")
```

### **Gene Expression by Genotype**



A14. The median expression level for genotype G|G is significantly lower than A|A's, suggesting that the SNP does affect the gene expression of ORMDL3. However, their whiskers overlap substantially and further statistical analysis would further distinguish the two. Their medians are and IQRs are not overlapping but their

whiskers are, final conclusion: SNP most likely does not affect the gene expression of ORMDL3.