

# Introduction to Genome Informatics

## Section 1. Identifying genetic variants of interest

This information is looked up using the Reference SNP cluster IDs in Ensembl.

Q1. The Verlaan et al. paper mentioned 4 candidate SNPs that demonstrate significant association with childhood asthma: rs12936231, rs8067378, rs9303277, and rs7216389.

The genes and location can both be found in the `location` Ensembl browser tab.

Q2. These four variants overlap or effect three genes: ZPBP2, IKZF3, GSDMB.

Q3. Information about rs8067378: Location: Chromosome 17: 39,894,595-39,895,595 3 Alleles: A/C/G

Q4. Three downstream genes for rs8067378: ORMDL3\_204, ORMDL3\_202, ORMDL3\_201

Sample genotypes can be looked up back in the `Variant` tab in the `Sample genotypes` section under “Explore this variant”.

```
#Mexican Ancestry in Los Angeles, California CSV file
rs <- read.csv("rs8067378.csv")

head(table(t(rs)))
```

```
##
##      -      A|A      A|G ALL, AMR, MXL      G|A
##     128      22      21      64      12
##     G|G
##      9
```

Q5. The proportion of (G|G) asthma-associated, homozygous genotypes in this dataset is 9/64 (14%).

Every person in the dataset has a number associated with them. We can use the “search for a sample” field to search for this number directly if we know what person we are trying to find.

Q6. For example, searching the particular sample HG00109 gives a person from the GBR population group with the genotype (G/G).

## Section 2. Initial RNA-Seq Analysis

Now, we want to understand whether the SNP will affect gene expression.

```
#We will be working with RNA sequences from HG00109
seq1 <- "HG00109_1.fastq"
seq2 <- "HG00109_2.fastq"
```

Q7. There are 3863 sequences in a fastqsanger format file sized 757 kb.

Q8. The mean GC content of file 2 is about 53% and the sequence length is 50-75.

Q9. No trimming is required for this dataset. The per base sequence quality is  $\geq 30$  for all bases.

## Section 3. Mapping RNA-Seq reads to genome

Here, we will be using Tophat to map the RNA sequences to the human genome.

Q10. The most accepted hits are located in the regions chr17: 38,140,121 - 38,154,856 and chr17: 38,061,531 - 38,080,564

Q11. The RNA reads seem to be mapping the genes *PSMD3* and *ORMDL3*.

The Cufflinks tool allows us to calculate gene expression (aligns RNA with annotated genome to result in list of genes being expressed, as evidenced by their transcription into RNA).

```
gene <- read.table("C:/Users/rodeo/Documents/School/BIMM 143/R Projects/Class10/Galaxy20-[Cufflinks_on_
gene[ gene$V10 > 0, 1]
```

```
## [1] "tracking_id" "ZBPB2"          "GSDMB"          "GSDMA"          "ORMDL3"
## [6] "PSMD3"
```

Q12. The FPKM for ORMDL3 is 136853. Other genes with FPKM  $> 0$  are ZBPB2, GSDMB, GSDMA, and PSMD3.

## Section 4. Population Scale Analysis

```
#Import Population Genotypes
as <- read.table("C:/Users/rodeo/Documents/School/BIMM 143/R Projects/Class10/rs8067378_ENSG00000172057
table(as)
```

```
## , , exp = 6.67482
##
##      geno
## sample A/A A/G G/G
##  HG00096  0  0  0
##  HG00097  0  0  0
```

##	HG00099	0	0	0
##	HG00100	0	0	0
##	HG00101	0	0	0
##	HG00102	0	0	0
##	HG00103	0	0	0
##	HG00104	0	0	0
##	HG00105	0	0	0
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##	HG00110	0	0	0
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##	HG00174	0	0	0
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##	NA20783	0	0	0
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```

## NA20787 0 0 0
## NA20790 0 0 0
## NA20792 0 0 0
## NA20795 0 0 0
## NA20796 0 0 0
## NA20797 0 0 0
## NA20798 0 0 0
## NA20799 0 0 0
## NA20800 0 0 0
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## NA20812 0 0 0
## NA20813 0 0 0
## NA20814 0 0 0
## NA20815 0 0 0
## NA20816 0 0 0
## NA20819 0 0 0
## NA20826 0 0 0
## NA20828 0 0 0
##
## , , exp = 6.9439
##
##      geno
## sample A/A A/G G/G
## HG00096 0 0 0
## HG00097 0 0 0
## HG00099 0 0 0
## HG00100 0 0 0
## HG00101 0 0 0
## HG00102 0 0 0
## HG00103 0 0 0
## HG00104 0 0 0
## HG00105 0 0 0
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## HG00104 0 0 0
## HG00105 0 0 0
## HG00106 0 0 0
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## HG00116 0 0 0
## HG00117 0 0 0
## HG00118 0 0 0
## HG00119 0 0 0
## HG00120 0 0 0
## HG00121 0 0 0
## HG00122 0 0 0
## HG00123 0 0 0
## HG00124 0 0 0
## HG00125 0 0 0

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##	NA20812	0	0	0
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## HG00103 0 0 0
## HG00104 0 0 0
## HG00105 0 0 0
## HG00106 0 0 0
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## HG00143 0 0 0
## HG00145 0 0 0
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## HG00101  0  0  0
## HG00102  0  0  0
## HG00103  0  0  0
## HG00104  0  0  0
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## HG00142  0  0  0
## HG00143  0  0  0
## HG00145  0  0  0
## HG00146  0  0  0
## HG00148  0  0  0
## HG00149  0  0  0
## HG00150  0  0  0

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## NA20772 0 0 0
## NA20773 0 0 0
## NA20774 0 0 0
## NA20778 0 0 0
## NA20783 0 0 0
## NA20785 0 0 0
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## NA20787 0 0 0
## NA20790 0 0 0
## NA20792 0 0 0
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## NA20796 0 0 0
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## NA20798 0 0 0
## NA20799 0 0 0
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## NA20801 0 0 0
## NA20802 0 0 0
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## NA20811 0 0 0
## NA20812 0 0 0
## NA20813 0 0 0
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## NA20816 0 0 0
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## HG00099 0 0 0
## HG00100 0 0 0
## HG00101 0 0 0
## HG00102 0 0 0
## HG00103 0 0 0
## HG00104 0 0 0
## HG00105 0 0 0
## HG00106 0 0 0
## HG00108 0 0 0
## HG00109 0 0 0
## HG00110 0 0 0
## HG00111 0 0 0

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##	HG00281	0	0	0
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##	HG00285	0	0	0

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##	HG00309	0	0	0
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## NA20809 0 0 0
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## NA20811 0 0 0
## NA20812 0 0 0
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## NA20826 0 0 0
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##
## , , exp = 11.39643
##

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##	HG00111	0	0	0
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```

## NA20778 0 0 0
## NA20783 0 0 0
## NA20785 0 0 0
## NA20786 0 0 0
## NA20787 0 0 0
## NA20790 0 0 0
## NA20792 0 0 0
## NA20795 0 0 0
## NA20796 0 0 0
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## NA20826 0 0 0
## NA20828 0 0 0
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## HG00105 0 0 0
## HG00106 0 0 0
## HG00108 0 0 0
## HG00109 0 0 0
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## HG00111 0 0 0
## HG00112 0 0 0
## HG00114 0 0 0
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## NA20812 0 0 0
## NA20813 0 0 0
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## NA20828 0 0 0
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## HG00100 0 0 0
## HG00101 0 0 0
## HG00102 0 0 0
## HG00103 0 0 0
## HG00104 0 0 0
## HG00105 0 0 0
## HG00106 0 0 0
## HG00108 0 0 0
## HG00109 0 0 0
## HG00110 0 0 0
## HG00111 0 0 0
## HG00112 0 0 0
## HG00114 0 0 0
## HG00115 0 0 0
## HG00116 0 0 0
## HG00117 0 0 0
## HG00118 0 0 0

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##	NA20804	0	0	0
##	NA20805	0	0	0
##	NA20806	0	0	0
##	NA20807	0	0	0
##	NA20808	0	0	0
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## NA20810    0    0    0
## NA20811    0    0    0
## NA20812    0    0    0
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## NA20814    0    0    0
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## NA20828    0    0    0
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## HG00100    0    0    0
## HG00101    0    0    0
## HG00102    0    0    0
## HG00103    0    0    0
## HG00104    0    0    0
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## HG00131    0    0    0
## HG00132    0    0    0
## HG00133    0    0    0
## HG00134    0    0    0
## HG00135    0    0    0
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## HG00137    0    0    0

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##	HG00259	0	0	0
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```

## NA20761 0 0 0
## NA20765 0 0 0
## NA20766 0 0 0
## NA20768 0 0 0
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## HG00111 0 0 0
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## NA20783 0 0 0
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##	NA20772	0	0	0
##	NA20773	0	0	0
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##	NA20783	0	0	0
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##	NA20808	0	0	0

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## NA20809    0    0    0
## NA20810    0    0    0
## NA20811    0    0    0
## NA20812    0    0    0
## NA20813    0    0    0
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## NA20815    0    0    0
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## NA20828    0    0    0
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## HG00100    0    0    0
## HG00101    0    0    0
## HG00102    0    0    0
## HG00103    0    0    0
## HG00104    0    0    0
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## HG00119    0    0    0
## HG00120    0    0    0
## HG00121    0    0    0
## HG00122    0    0    0
## HG00123    0    0    0
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## HG00125    0    0    0
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## HG00127    0    0    0
## HG00128    0    0    0
## HG00129    0    0    0
## HG00130    0    0    0
## HG00131    0    0    0
## HG00132    0    0    0
## HG00133    0    0    0
## HG00134    0    0    0
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## HG00136    0    0    0

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##	NA20756	0	0	0

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## NA20757 0 0 0
## NA20758 0 0 0
## NA20759 0 0 0
## NA20760 0 0 0
## NA20761 0 0 0
## NA20765 0 0 0
## NA20766 0 0 0
## NA20768 0 0 0
## NA20769 0 0 0
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## NA20812 0 0 0
## NA20813 0 0 0
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## HG00099 0 0 0

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##
## geno

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## NA20792 0 0 0
## NA20795 0 0 0
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## NA20797 0 0 0
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## NA20800 0 0 0
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## NA20798 0 0 0
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## NA20809 0 0 0
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## NA20812 0 0 0
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## HG00111 0 0 0
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## HG00119 0 0 0
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## HG00124 0 0 0
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## HG00145 0 0 0
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## HG00105 0 0 0
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## HG00119 0 0 0
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## HG00126 0 0 0
## HG00127 0 0 0
## HG00128 0 0 0

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##	NA20582	0	0	0
##	NA20585	0	0	0
##	NA20586	0	0	0

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## NA20752 0 0 0
## NA20754 0 0 0
## NA20756 0 0 0
## NA20757 0 0 0
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## NA20770 0 0 0
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## NA20772 0 0 0
## NA20773 0 0 0
## NA20774 0 0 0
## NA20778 0 0 0
## NA20783 0 0 0
## NA20785 0 0 0
## NA20786 0 0 0
## NA20787 0 0 0
## NA20790 0 0 0
## NA20792 0 0 0
## NA20795 0 0 0
## NA20796 0 0 0
## NA20797 0 0 0
## NA20798 0 0 0
## NA20799 0 0 0
## NA20800 0 0 0
## NA20801 0 0 0
## NA20802 0 0 0
## NA20803 0 0 0
## NA20804 0 0 0
## NA20805 0 0 0
## NA20806 0 0 0
## NA20807 0 0 0
## NA20808 0 0 0
## NA20809 0 0 0
## NA20810 0 0 0
## NA20811 0 0 0
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## NA20816 0 0 0
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## NA20826 0 0 0
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##
## , , exp = 17.46326
##

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##	HG00105	0	0	0
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##	HG00108	0	0	0
##	HG00109	0	0	0
##	HG00110	0	0	0
##	HG00111	0	0	0
##	HG00112	0	0	0
##	HG00114	0	0	0
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##	NA20772	0	0	0
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##	NA20774	0	0	0

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## NA20778 0 0 0
## NA20783 0 0 0
## NA20785 0 0 0
## NA20786 0 0 0
## NA20787 0 0 0
## NA20790 0 0 0
## NA20792 0 0 0
## NA20795 0 0 0
## NA20796 0 0 0
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## NA20809 0 0 0
## NA20810 0 0 0
## NA20811 0 0 0
## NA20812 0 0 0
## NA20813 0 0 0
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## HG00103 0 0 0
## HG00104 0 0 0
## HG00105 0 0 0
## HG00106 0 0 0
## HG00108 0 0 0
## HG00109 0 0 0
## HG00110 0 0 0
## HG00111 0 0 0
## HG00112 0 0 0
## HG00114 0 0 0
## HG00115 0 0 0

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##	HG00118	0	0	0
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##	HG00189	0	0	0
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## HG00116 0 0 0
## HG00117 0 0 0
## HG00118 0 0 0

```

```

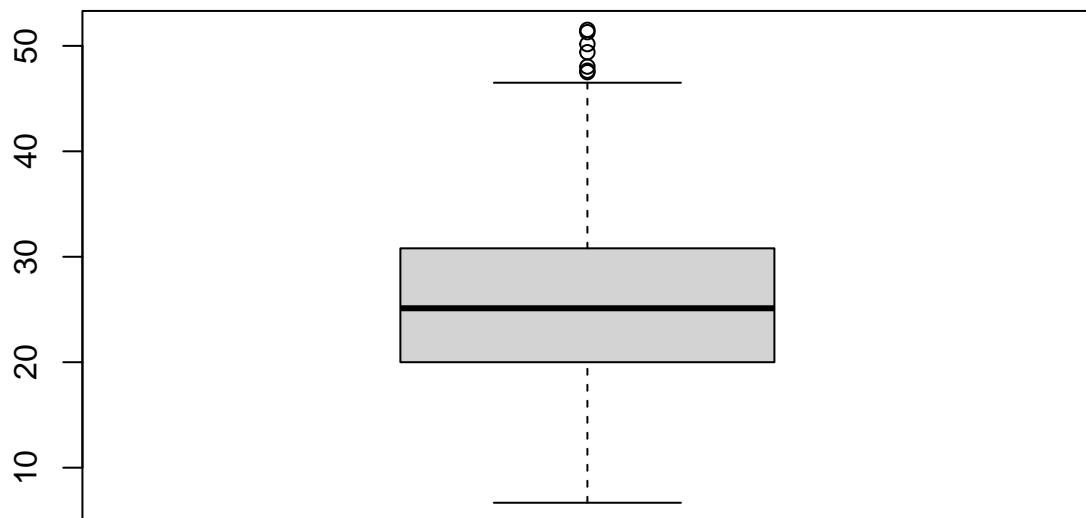
## HG00119 0 0 0
## HG00120 0 0 0
## HG00121 0 0 0
## HG00122 0 0 0
## HG00123 0 0 0
## HG00124 0 0 0
## HG00125 0 0 0
## HG00126 0 0 0
## HG00127 0 0 0
## HG00128 0 0 0
## HG00129 0 0 0
## HG00130 0 0 0
## HG00131 0 0 0
## HG00132 0 0 0
## HG00133 0 0 0
## HG00134 0 0 0
## HG00135 0 0 0
## HG00136 0 0 0
## HG00137 0 0 0
## HG00138 0 0 0
## HG00139 0 0 0
## HG00141 0 0 0
## HG00142 0 0 0
## HG00143 0 0 0
## HG00145 0 0 0
## HG00146 0 0 0
## HG00148 0 0 0
## HG00149 0 0 0
## HG00150 0 0 0
## HG00151 0 0 0
## HG00152 0 0 0
## HG00154 0 0 0
## HG00155 0 0 0
## HG00156 0 0 0
## HG00157 0 0 0
## HG00158 0 0 0
## HG00159 0 0 0
## HG00160 0 0 0
## HG00171 0 0 0
## HG00173 0 0 0
## HG00174 0 0 0
## HG00176 0 0 0
## HG00177 0 0 0
## HG00178 0 0 0
## HG00179 0 0 0
## HG00180 0 0 0
## HG00181 0 0 0
## HG00182 0 0 0
## HG00183 0 0 0
##
## [ reached getOption("max.print") -- omitted 393 row(s) and 389 matrix slice(s) ]

```

```
summary(as)
```

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

```
boxplot(as$exp, group = factor(as$geno))
```



Q13.