### 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 dowstream genes for rs8067378: ORMDL3\_204, ORMDL3\_202, ORMDL3\_201

Sample genotypes can be looked up back in the Variant tab in the Sameple 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</pre>
```

Q5. The proportion of (G|G) as thma-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, serching the particular sample HG00109 gives a person from the GBR population group with the genotype (G/G).

# Section 2. Initial RNA-Seq Analysis

##

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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"</pre>
```

- Q7. There are 3863 sequences in a fastqsanger format file sized 757 kb.
- Q8. The measn 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 >=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" "ZPBP2" "GSDMB" "GSDMA" "ORMDL3"
## [6] "PSMD3"
```

Q12. The FPKM for ORMDL3 is 136853. Other genes with FPKM > 0 are ZPBP2, GSDMB, GSDMA, and PSMD3.

### Section 4. Population Scale Analysis

A/A A/G G/G

0

0

## sample

HG00096

HG00097

##

##

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

## , , exp = 6.67482
## geno</pre>
```

##	HG00099	0	0	0
##	HG00100	0	0	0
##	HG00101	0	0	0
##	HG00102	0	0	0
##	HG00103	0	0	0
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##	NA20506	0	0	0
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## NA20792 0 0 0					
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## NA12283	##	NA12275	0	0	0
## NA12286	##	NA12282	0	0	0
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## NA12383	##	NA12347	0	0	0
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	NA19095	0	0	0
## ##	NA19096		_	_
## ##	NA19098 NA19099	0	0	0
## ##	NA19099 NA19102	0	0	0
## ##	NA19102 NA19107	0	0	0
## ##	NA19107 NA19108	0	0	0
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## ##	NA19113 NA19114	0	0	0
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## ##	NA19117 NA19118	0		0
## ##	NA19118 NA19119	0	0	0
## ##	NA19119 NA19121	0		0
## ##	NA19121 NA19129	0	0	0
## ##	NA19129 NA19130	0	0	0
## ##	NA19130 NA19131	0	0	0
## ##	NA19131 NA19137	0	0	0
## ##	NA19137 NA19138	0	0	0
##	NATATOR	U	U	U

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##	NA19144	0	0	0
##	NA19146	0	0	0
##	NA19147	0	0	0
##	NA19149	0	0	0
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##	NA19153	0	0	0
##	NA19159	0	0	0
##	NA19160	0	0	0
##	NA19171	0	0	0
##	NA19172	0	0	0
##	NA19175	0	0	0
##	NA19184	0	0	0
##	NA19185	0	0	0
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##	NA19190	0	0	0
##	NA19197	0	0	0
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## ##	NA19200 NA19201	0	0	0
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## NA20774 0 0 0 0 ## NA20778 0 0 0 0 ## NA20785 0 0 0 0 ## NA20786 0 0 0 0 ## NA20797 0 0 0 0 ## NA20797 0 0 0 0 ## NA20797 0 0 0 0 0 ## NA20807 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA20772		0	1
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## NA20797 0 0 0 0 ## NA20798 0 0 0 0 ## NA20799 0 0 0 0 ## NA20800 0 0 0 0 ## NA20802 0 0 0 0 ## NA20805 0 0 0 0 ## NA20805 0 0 0 0 ## NA20806 0 0 0 0 ## NA20808 0 0 0 0 ## NA20808 0 0 0 0 ## NA20808 0 0 0 0 ## NA20811 0 0 0 0 ## NA20812 0 0 0 ## NA20813 0 0 0 ## NA20814 0 0 0 ## NA20815 0 0 0 ## NA20816 0 0 0 ## NA20819 0 0 0	##	NA20795			
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## NA20804 0 0 0 0 ## NA20805 0 0 0 0 ## NA20806 0 0 0 0 ## NA20808 0 0 0 0 ## NA20810 0 0 0 ## NA20811 0 0 0 ## NA20813 0 0 0 ## NA20814 0 0 0 ## NA20815 0 0 0 ## NA20816 0 0 0 ## NA20819 0 0 0 0 ## NA20819 0 0 0	##				
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## NA20816 0 0 0 ## NA20819 0 0 0					
## NA20819 0 0 0					
## NA20826 0 0 0					
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## sample
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## NA11918 0 0 0 0   ## NA11930 0 0 0 0   ## NA11931 0 0 0 0   ## NA11992 0 0 0 0   ## NA11993 0 0 0 0   ## NA11994 0 0 0 0   ## NA11995 0 0 0 0   ## NA12004 0 0 0 0   ## NA12005 0 0 0 0   ## NA12006 0 0 0 0   ## NA12043 0 0 0 0   ## NA12044 0 0 0 0   ## NA12045 0 0 0 0   ## NA12045 0 0 0 0   ## NA12045 0 0 0 0   ## NA12156 0 0 0 0   ## NA12156 0 0 0 0   ## NA12234 0 0 0 0   ## NA12249 0 0 0 0   ## NA12340 0 0 0   ## NA12341 0 0 0 0   ## NA12341	##	NA11893	0	0	0
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## NA11994 0 0 0 0 ## NA12004 0 0 0 ## NA12005 0 0 0 0 ## NA12006 0 0 0 0 ## NA12043 0 0 0 0 ## NA12045 0 0 0 0 ## NA12045 0 0 0 0 ## NA12155 0 0 0 0 ## NA12234 0 0 0 0 ## NA12275 0 0 0 0 ## NA12286 0 0 0 0 ## NA12287 0 0 0 0 ## NA12341 0 0 0 0 ## NA12341 0 0 0 0 ## NA12342 0 0 0 0 ## NA12343 0 0 0 0 ## NA12340 0 0 0 0 ## NA12341 0 0 0 0 0 ## NA12341 0 0 0 0 0 ## NA12342 0 0 0 0 0 ## NA12343 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA11992	0	0	0
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## NA11920 0 0 0 0 ## NA11931 0 0 0 0 ## NA11992 0 0 0 0 ## NA11993 0 0 0 0 0 ## NA11994 0 0 0 0 ## NA12004 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA11894	0	0	0
## NA11930 0 0 0 0 ## NA11931 0 0 0 0 ## NA11992 0 0 0 0 ## NA11993 0 0 0 0 ## NA11995 0 0 0 0 ## NA12005 0 0 0 0 0 ## NA12045 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA11918			
## NA11931 0 0 0 0 ## NA11992 0 0 0 0 ## NA11993 0 0 0 0 ## NA11995 0 0 0 0 ## NA12004 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA11920			
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##
     HG00097
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                         0
##
     HG00099
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## NA20543 0 0 0 ## NA20544 0 0 0	##	NA20541	0	0	0
## NA20544 0 0 0	##	NA20542	0	0	0
	##	NA20543	0	0	0
## NA20581 0 0 0	##	NA20544	0	0	0
	##	NA20581	0	0	0

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## NA20797 0 0 0 0 ## NA20798 0 0 0 0 ## NA20799 0 0 0 0 0 ## NA20800 0 0 0 0 0 ## NA20802 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA20795	0	0	0
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## NA12273 0 0 0 0 ## NA12275 0 0 0 0 ## NA12282 0 0 0 0 ## NA12283 0 0 0 ## NA12286 0 0 0 ## NA12287 0 0 0 ## NA12340 0 0 0 ## NA12341 0 0 0 ## NA12342 0 0 0 ## NA12342 0 0 0 ## NA12343 0 0 0 ## NA12348 0 0 0 ## NA12348 0 0 0 ## NA12348 0 0 0 ## NA12349 0 0 0 ## NA12400 0 0 0 ## NA12413 0 0 0 ## NA12413 0 0 0 ## NA12716 0 0 0 ## NA12718 0 0 0 ## NA12718 0 0 0 ## NA12750 0 0 0 ## NA12760 0 0 0 ## NA12775 0 0 0 ## NA12775 0 0 0 ## NA12776 0 0 0 ## NA12777 0 0 0 ## NA12778 0 0 0 ## NA12778 0 0 0 ## NA12813 0 0 0 ## NA12814 0 0 0 ## NA12815 0 0 0 ## NA12815 0 0 0 ## NA12816 0 0 0 ## NA12817 0 0 0 ## NA12818 0 0 0 ## NA12840 0 0 0 ## NA18480 0 0 0 ## NA18480 0 0 0 ## NA18480 0 0 0 ## NA18490 0 0 0	##	NA12249	0	0	0
## NA12275	##	NA12272	0	0	0
## NA12282 0 0 0 0 ## NA12283 0 0 0 0 ## NA12286 0 0 0 0 ## NA12287 0 0 0 0 ## NA12340 0 0 0 0 ## NA12341 0 0 0 0 ## NA12342 0 0 0 0 ## NA12348 0 0 0 0 ## NA12348 0 0 0 0 ## NA12348 0 0 0 0 ## NA12430 0 0 0 0 ## NA12430 0 0 0 0 ## NA12717 0 0 0 0 ## NA12717 0 0 0 0 ## NA12718 0 0 0 0 ## NA12718 0 0 0 0 ## NA12718 0 0 0 0 ## NA12719 0 0 0 0 ## NA12719 0 0 0 0 ## NA12719 0 0 0 0 0 ## NA12719 0 0 0 0 0 ## NA12719 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA12273	0	0	0
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## NA12286	##	NA12282	0	0	0
## NA12287 0 0 0 0 ## NA12340 0 0 0 0 ## NA12341 0 0 0 0 0 ## NA12342 0 0 0 0 0 ## NA12342 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA12283	0	0	0
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## NA12778 0 0 0 0 ## NA12812 0 0 0 0 ## NA12813 0 0 0 0 ## NA12815 0 0 0 0 ## NA12827 0 0 0 0 ## NA12829 0 0 0 0 ## NA12830 0 0 0 0 ## NA12842 0 0 0 0 ## NA12872 0 0 0 0 ## NA12874 0 0 0 0 ## NA12874 0 0 0 0 ## NA12889 0 0 0 0 ## NA12890 0 0 0 ## NA12890 0 0 0 ## NA18486 0 0 0 ## NA18488 0 0 0 0 ## NA18488 0 0 0 0 ## NA18489 0 0 0 0 ## NA18499 0 0 0 0 ## NA18499 0 0 0 0 ## NA18499 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	##	NA12776	0	0	0
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## NA12842 0 0 0 0 ## NA12843 0 0 0 0 ## NA12872 0 0 0 0 ## NA12873 0 0 0 0 ## NA12874 0 0 0 0 ## NA12889 0 0 0 0 ## NA18486 0 0 0 0 ## NA18488 0 0 0 0 ## NA18489 0 0 0 ## NA18499 0 0 0 0 ## NA18499 0 0 0 0	##				
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## NA18487 0 0 0 ## NA18488 0 0 0 ## NA18489 0 0 0 ## NA18498 0 0 0 ## NA18499 0 0 0	##				
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## NA18498 0 0 0 ## NA18499 0 0 0	##				
## NA18499 0 0 0	##				
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## NA18502 0 0 0	##				
	##	NA18502	0	0	0

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##	NA18505	0	0	0
##	NA18508			
##	NA18510	0	0	0
##	NA18511	0	0	0
##	NA18517			
##	NA18519	0	0	0
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##	NA18909	0	0	0
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##	NA19213	0	0	0
##	NA19214	0	0	0
##	NA19222	0	0	0
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##	NA19225	0	0	0
##	NA19235	0	0	0
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## ##	NA20507	0	0	0
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## ##	NA20509 NA20510	0	0	0
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##	NA20512	U	0	0

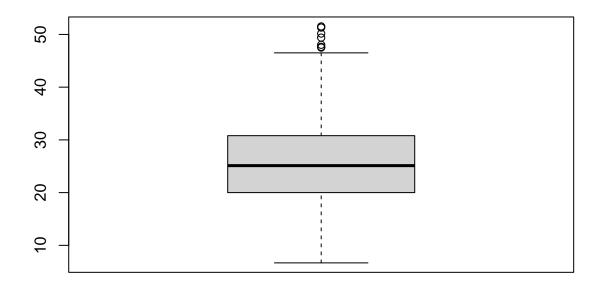
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##
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     HG00097
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##
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##
     HG00099
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     HG00100
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##
     HG00101
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     HG00102
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     HG00104
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     HG00105
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     HG00112
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     HG00117
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     HG00118
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HG00119
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##
    [ 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.