

# Class 12: Genome Informatics

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## Section 1: Proportion of G/G in population

Downloaded CSV file from Ensembl < [https://useast.ensembl.org/Homo\\_sapiens/Variation/Sample?db=core;v=rs8067378;vdb=variation;vf=959672880#373531\\_tablePanel](https://useast.ensembl.org/Homo_sapiens/Variation/Sample?db=core;v=rs8067378;vdb=variation;vf=959672880#373531_tablePanel) > Here we read this CSV file

```
mxl <- read.csv("373531-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
head(mxl)
```

	Sample..Male.Female.Unknown.	Genotype..forward.strand.	Population.s.	Father
1	NA19648 (F)	A A	ALL, AMR, MXL	-
2	NA19649 (M)	G G	ALL, AMR, MXL	-
3	NA19651 (F)	A A	ALL, AMR, MXL	-
4	NA19652 (M)	G G	ALL, AMR, MXL	-
5	NA19654 (F)	G G	ALL, AMR, MXL	-
6	NA19655 (M)	A G	ALL, AMR, MXL	-
Mother				
1	-			
2	-			
3	-			
4	-			
5	-			
6	-			

```
table(mxl$Genotype..forward.strand.)
```

A A	A G	G A	G G
22	21	12	9

```
table(mx1$Genotype..forward.strand.) / nrow(mx1)
```

A A	A G	G A	G G
0.343750	0.328125	0.187500	0.140625

Let's look at a different population. I picked the GBR.

```
gbr <- read.csv("373522-SampleGenotypes-Homo_sapiens_Variation_Sample_rs8067378.csv")
table(gbr$Genotype..forward.strand.) / nrow(gbr)
```

A A	A G	G A	G G
0.2527473	0.1868132	0.2637363	0.2967033

This variant that is associated with childhood asthma is more frequent in the GBR population than the MXL population.

Let's look into this further.

##Section 4: Population Scale Analysis One sample is obviously not enough to know what is happening in a population. You are interested in assessing genetic differences on a population scale.

So, you processed about ~230 samples and did the normalization on a genome level. Now, you want to find whether there is any association of the 4 asthma-associated SNPs (rs8067378...) on ORMDL3 expression.

How many samples do we have?

```
expr <- read.table("rs8067378_ENSG00000172057.6.txt")
head(expr)
```

	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

```
nrow(expr)
```

```
[1] 462
```

```
table(expr$geno)
```

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

```
library(ggplot2)
```

Let's make a boxplot with this data

```
ggplot(expr) + aes(x = geno, y = exp, fill = geno) + geom_boxplot(notch = T)
```

