

GWAS Interpretation
By: Alicia Martin

From the class GWAS with data combined between last year and this year, we have the following data for the bitter phenotype with the most significantly correlated SNP ($p=6e-5$, $N=58$)

```
bitter      x713598
              CC CG GG NULL
bitter_no   9  4  1   1
bitter_yes  1 29 14   1
```

From the genotypes, we can construct the following contingency table:

		Can taste bitter?	
		Yes	No
Allele Counts	C	31	22
	G	57	6

From this contingency table, we can calculate an odds ratio and likelihood ratio.

Odds Ratio

Definition: odds ratio – a measure of effect size, describing the strength of association or non-independence between two binary data values.

In a case control study, this is the ratio between the fraction with the risk variant versus non-risk variant in the groups of affected versus the controls, i.e. expressed in terms of probabilities conditional on the affection status:

OR = $(a \times d)/(b \times c)$ when you have the following contingency table:

		Treatment	
		Placebo	Drug
Event	Occurs	a	b
	Does not occur	c	d

Therefore, the OR is:

$$\text{OR} = (31 * 6) / (22 * 57)$$

$$\text{OR} = 0.15$$

Likelihood Ratio

Given a risk allele, what is the likelihood of having the disease?

$$\text{LR} = P(A \mid \text{Trait}) / P(A \mid \text{No trait})$$
$$P(A \mid \text{Trait}) = P(\text{Trait} \ \& \ A) / P(\text{Trait})$$

In our example:

$$P(G \mid \text{bitter taste}) = 57 / (57 + 31)$$
$$P(G \mid \text{No trait}) = 6 / (6 + 22)$$

$$\text{LR} = [57 / (57 + 31)] / [6 / (6 + 22)]$$
$$\text{LR} = 3.02$$

This is the likelihood of tasting bitter given a G allele. What does this mean in terms of GG and GC? You can use $P(\text{GG} \mid \text{Trait}) / P(\text{GG} \mid \text{No trait})$ and look at GG versus GC and CC using the genotypes table from last week.

Relative Risk

The relative risk (RR) is the probability that a member of an exposed group will develop a disease relative to the probability that a member of an unexposed group will develop that same disease. This calculation requires that we know something about the overall incidence of the trait.

$$R_{CC} = P(\text{trait} \mid CC) / \text{population prevalence of trait}$$
$$R_{CC} = P(\text{affected} \mid CC \text{ genotype}) / \text{population affected prevalence}$$