

GWAS Interpretation  
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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?

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

$$P(A \mid \text{Trait}) = P(\text{Trait} \& A) / P(\text{Trait})$$

In our example:

$$P(G \mid \text{bitter taste}) = 57 / (57 + 31)$$

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

$$LR = [57 / (57 + 31)] / [6 / (6 + 22)]$$

$$LR = 3.02$$

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

### Relative Risk

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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.

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

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