

# Genetics Review

## Terms:

The main terms have been identified in the online quiz. Here is a good list.

- |                 |                          |                           |
|-----------------|--------------------------|---------------------------|
| 1. Gene         | 6. Genotype              | 11. Pleiotropic           |
| 2. Locus        | 7. Phenotype             | 12. Multi-gene trait      |
| 3. Allele       | 8. Dominant              | 13. epistatic/epistasis   |
| 4. Heterozygous | 9. Recessive             | 14. true-breeding (see 5) |
| 5. Homozygous   | 10. incomplete dominance | 15. trisomy               |

## The idea of a gene:

How are the various ways "gene" has been defined? We have used either "unit of inheritance;" "a stretch of DNA that encodes a protein and its regulatory sequences" most of the time. But, what about genes that encode tRNAs or the small nuclear RNAs? They don't encode proteins, but certainly could have alleles that result in phenotypes. So, gene could be any DNA that results in a phenotype that is inherited. Why are some alleles "recessive" and others "dominant?"

## Crosses

Data will be similar to those seen in VGL (but maybe not identical). Questions like the online quiz in which you need to do a Punnett square to answer a multiple choice will be there. You should be able recognize in a cross whether a trait is dominant, recessive or shows incomplete dominance. You should also recognize sex-linked. To do this, you should know the sorts of ratios expected for a cross (1:2:1 for genotype, 3:1 for phenotype if simple dominance; 1:2:1 for both genotype *and* phenotype if incomplete dominance). Examples:

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### Incomplete dominance:

True-breeding Red flower by true-breeding white flower gives all pink flowers in the F1. F1 cross depicted below:

The RR is red, ww is white, RW is pink (1:2:1)

Neither allele is dominant

Reds and whites are each always true-breeding

Pinks give 1:2:1 when bred to each other

Red by pink gives 50% each RR (red) and RW (pink)

Same for white by pink.

If, say, red were dominant,  $\frac{3}{4}$  of the flowers would be red.

	R	W
R	RR	RW
W	RW	WW

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### Co-dominance:

Really closely related to incomplete dominance and kind of a semantic difference. It's when both traits are seen. A/B blood type could fit this. But, stick with incomplete dominance most of the time.

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### X-linked:

The small sex chromosome *does not* have the same genes as the large. So, our "Y" chromosome will not have alleles that can cover up mutant recessive alleles. Since males only

have one X (or females only have one "W" in birds), Recessive traits are seen far more often in the "heterogametic" sex (XY in humans). Also, for sex-linked traits, it matters whether the mother or father has the trait in question. Males get their lone X from their mother. Consider white-eyed male by red-eyed female fly: The F1 looks normal, with white-eyes being recessive. If I had done it with a white-eyed female and a red-eyed male, I would get:

<b>Red male X white female</b>	$X^r$	Y
$X^w$	$X^rX^w$	$X^wY$
$X^w$	$X^rX^w$	$X^wY$
<b>white male X red female</b>	$X^w$	Y
$X^r$	$X^rX^w$	$X^rY$
$X^r$	$X^rX^w$	$X^rY$

All the males are white-eyed.

All the females are red-eyed heterozygotes.

You might want to practice filling out the squares for the expected F1 cross for each of these cages ( $X^rX^w$  by  $X^rY$  and  $X^rX^w$  by  $X^wY$ ).

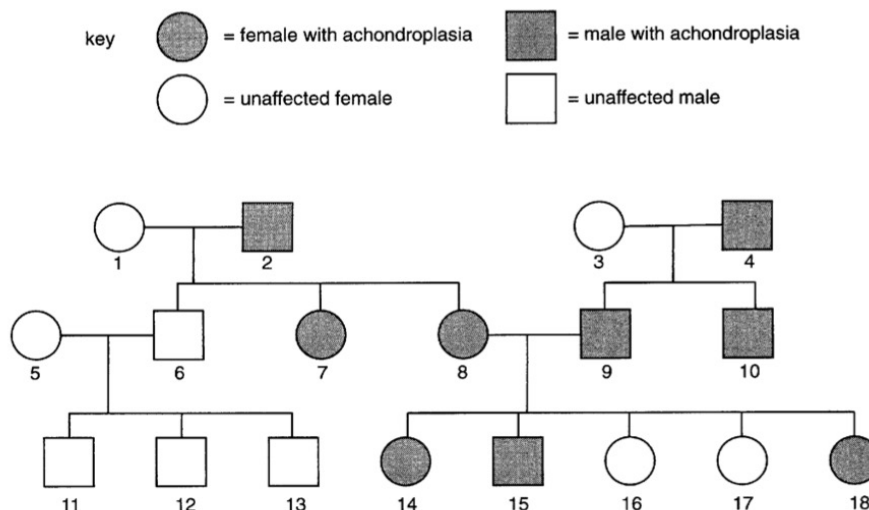
## Dihybrid:

You should be able to recognize the patterns expected for both a dihybrid (9:3:3:1) and test cross (1:1:1:1) and perform a chi-square to assess if there is likely linkage (test the null hypothesis that any deviation is random fluctuation). Equations will be provided. The blog has examples of these.

DON'T BE SHOCKED if a cross combines components of these (eg, a sex link in a dihybrid cross).

## Pedigree:

They will be simple. The key thing to look for is any parents that have one trait ("A") and have offspring with the other trait ("B"). B is therefore recessive since it was covered up in the parents. It doesn't matter whether "A" or "B" were considered the mutant trait (or "affected"). Below is a pedigree for achondroplasia (a form of dwarfism). Look at individuals 8



and 9. They are both dwarfs and have unaffected offspring (16 and 17). Thus, this form of dwarfism is dominant and individuals 8&9 are heterozygous.

For X-linked, you see a recessive show up far more in males, as before. But, you never see passage from father to son, as you would for a very rare Y-linked.

### **Karyotype:**

Again, simple. Look for trisomy (three copies of one chromosome). Look for whether the individual is XY or XX. The chromosomes will be aligned for you, if there is a karyotype.

### **Chi-squared**

You will be given example data and all the equations necessary. You should know how to write a null hypothesis (i.e. "This is just random fluctuations from the expected pattern") and test whether the data are a good fit for that hypothesis. Be sure you know what you can and **cannot** claim based on your result.

### **Transcription/Translation:**

There will be no fact-based questions such as the matching columns etc. However, there may be questions that include information about a mutation that pertains to transcription and translation. For example, if I included information about the mutation that causes blue eyes (it is a mutation to the promoter of a melanin transport protein), I might ask you about how that could affect the phenotype etc. So, I recommend you think about:

- What would happen if there were a mutation to a splice site in a gene?
- What would happen if a promoter mutation resulted in expression of a protein in the wrong organ? Or, failed to work in an organ where it should?
- With respect to pleiotropy—mutations that have many, varied effects—one class could include a mutation in the gene for a transcription factor that results in many genes being improperly regulated.