Notes

Biology 102
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Chapter 12

Reading: Chapter 12

Topic: Patterns of Inheritance

Important vocabulary terms used throughout the genetics unit:

- **Chromosome**: Strands of DNA in the nucleus of the cell. Technically, it is a chromosome only when it is wound up around special histone proteins just before cell division. However, it is convenient for us to refer to "chromosomes" any time we discuss a DNA strand that carries genes. Humans have 23 pairs of DNA strands, and therefore 23 pairs of chromosomes (a total of 46).
- **Gene**: A segment of DNA that codes for a particular protein, and therefore a particular trait.
- **Allele**: A "version" of a gene. Many genes come in two "versions": dominant and recessive. Some come in multiple "versions," such as the A, B, and O alleles for blood type.
- **Phenotype**: The outward expression of a gene; that is, the trait itself.
- **Genotype**: The actual genes that code for a particular phenotype. We often represent the genes with letters.
- **Dominant**: An allele that is expressed if a person inherits at least one copy of that allele from one parent. "Dominant" in genetics has nothing to do with how prevalent a trait is in a population (the term for that is "frequency" of an allele). Dominant alleles are often represented using a capital letter.
- **Recessive**: An allele that is expresses itself in the phenotype only if two copies of the recessive allele are inherited, one from each parent. We usually represent recessive alleles using a small-case letter.
- **Homozygous**: A genotype in which a person inherits the same allele for a gene from each parent; that is, a person is homozygous dominant if they receive two copies of the dominant allele, one from each parent (written as AA). Likewise, a person is homozygous recessive if they inherit two copies of the recessive allele, one from each parent (written as aa).
- **Heterozygous**: A genotype in which a person inherits one dominant allele from one parent and one recessive allele from the other parent (written as Aa).
- **Diploid**: A cell which has two copies of each chromosome (the normal state of cells, since we all get one copy of each chromosome from Mom and one copy from Dad).
- **Haploid**: A cell which has only one copy of each chromosome. In animals, sex cells are haploid.

Main concepts:

- Gregor Mendel was the first to propose and gather the data to support the concept that inheritance was predictable, and controlled by individual factors that we now call genes (Mendel called them "elementen").
- •Mendel worked with traits in peas that we now know are single-gene traits carried on separate chromosomes (there is some evidence that Mendel was on the track of discovering linkage — multiple genes on the same chromosome — before his work was interrupted).
- Mendel developed three laws of heredity:
  - **Law of Dominance**: The traits that Mendel studied were single-gene traits coded for by alleles that were either dominant or recessive. The Law of Dominance states that traits are controlled by pairs of physical units, which we call genes. The genes come in two types: the dominant allele and the recessive allele. If an offspring inherits at least one copy of the dominant allele, it will show the dominant phenotype (the recessive allele may be present, but its effects are masked). Two true-breeding parents, one with the dominant phenotype and one with the recessive phenotype, will always produce offspring with the dominant phenotype, but those offspring will be heterozygous (will carry the recessive allele in their genotype).
  - **Law of Segregation**: During meiosis (production of sex cells), pairs of alleles are separated from one another, so that only one copy of each gene goes into each sex cell. A heterozygous individual (Aa) can put either the A allele (dominant allele) or the a allele (recessive) allele into its sex cells. Likewise, a homozygous dominant individual (AA) can put one A allele into its sex cells, and a homozygous recessive individual (aa) can put one a allele into its sex cells.
  - **Law of Independent Assortment**: During meiosis (sex cell production), the alleles sort independently of each other. That is, if an individual is heterozygous for two traits (Aa and Bb), if the dominant allele for the first trait (A) ends up in one sex cell, the dominant allele for the second trait (B) doesn't necessarily follow. By this law, an AaBb individual could produce sex cells carrying AB, Ab, aB, or ab.

- Punnett squares (figure 12-11a) are useful tools for predicting the possible outcomes of any one cross. When writing a Punnett square, the alleles that one parent can donate go across the top of the square, and the alleles the other parent can donate go on one side of the square. A Punnett square makes predictions about probability. It does not depict a guaranteed actual outcome. Probabilities can also be calculated mathematically, as Mendel did (see figure 12-11b).
- Incomplete dominance was discovered after Mendel's work. In incomplete dominance, both alleles are expressed in the phenotype of the heterozygous individuals. An example is in snapdragons, which some in red (RR), white (R'R') and pink (RR'). Notice that instead of capital and small-case letters, we use capital letters with a ' ("prime") for the two alleles, since neither is dominant over the other. (Co-dominance is very similar, except that the two traits can be seen in individually in the phenotype instead of appearing to be blended. An example is a "roan" horse, which has both red and white hairs.)
- Multiple traits may be inherited on different chromosomes. When traits are carried on different chromosomes, they sort independently of one another during gamete formation; that is, if a person is heterozygous for traits A and B (genotype Aa Bb), donating a dominant allele A does not necessarily mean that the dominant allele B will also go into that same gamete. This illustrates Mendel's Law of Independent Assortment.
- The probability of any one offspring inheriting any particular combination of traits on different chromosomes can be determined using a Punnett square for a dihybrid cross. Figure 12-14 shows a dihybrid cross Punnett square. Notice that two letters (symbols for alleles) are in each of the gametes drawn across the top and down the side of the square. The individual can donate *one of each kind of chromosome* in each gamete. If we want to track two traits at once, we must first determine how the traits sort out in the gametes.
- If two individuals who are heterozygous for two traits carried on separate chromosomes (Aa Bb) are crossed, the probable ratios of phenotypes in the offspring are:
    - 9 dominant for both traits
    - 3 dominant for trait A, recessive for trait B
    - 3 recessive for trait A, dominant for trait B
    - 1 recessive for both traits
- Here is an example for height in peas (tall vs. short) and pea color (yellow vs. green):

	TY	Ty	tY	ty
TY	TTYY	TTYy	TtYY	TtYy
Ty	TTYy	TTyy	TtYy	Ttyy
tY	TtYY	TtYy	ttYY	ttYy
ty	ttYy	Ttyy	ttYy	ttyy

Notice the ratios:
- 9/16 tall, yellow
- 3/16 tall, green
- 3/16 short, yellow
- 1/16 short, green

Compare this square to the one in figure 12-14.

- Before Punnett squares, Mendel determined probabilities for a dihybrid cross mathematically as follows:
    - Start by determining the probabilities of inheriting the two traits independently. In this example, we're using pea color (yellow vs. green) and pea flower color (purple vs. white).

pea color

	Y	y
Y	YY	Yy
y	Yy	yy

Yellow = 3/4 or 0.75

flower color

	P	p
P	PP	Pp
p	Pp	pp

Purple = 3/4 or 0.75

green = 1/4 or 0.25 white = 1/4 or 0.25

- Multiply the probabilities of the two outcomes you want to track to find the probability of both outcomes occurring together:
- Probability of yellow peas AND purple flowers = .75 x .75 = .56 (which equals the 9/16 ratio seen in the Punnett square)
- Probability of yellow peas AND white flowers = .75 x .25 = .19 (which equals the 3/16 ratio seen in the Punnett square)
- Probability of green peas AND purple flowers = .75 x .25 = .19 (which equals the 3/16 ratio seen in the Punnett square)
- Probability of green peas AND white flowers = = .25 x .25 = .06 (which equals the 1/16 ratio seen in the Punnett square)

Common misconceptions:

- Students often have trouble with the word “dominant” (often misspelled “dominate”) which has many meanings in everyday language, but has one very specific meaning in genetics. A dominant trait is one which shows in the phenotype even if only one allele for that trait is inherited. “Dominant” does NOT mean any of the following: strongest; most common in the population; taking over in the population; cancels out the recessive allele; suppresses the recessive allele; any trait that shows in the phenotype. Alleles are either dominant or not. They do not become “more dominant.”
- The most common errors that students make when drawing a dihybrid Punnett square is to see four squares across and four alleles (two pairs), and assume that they should put one letter (one allele) over each of the four boxes. However, this does not reflect the fact that each gamete has one copy of each kind of allele. The letters over the boxes must represent what is in the gametes.
- Students often believe the 9:3:3:1 ratio always holds true for dihybrid crosses, regardless of what the genotypes of the parents are.

Reading notes:

- Define: locus, gene, allele, homozygous, heterozygous, hybrid.
- State why inheritance in peas was a good model for Mendel to study.
- Define: parental generation (P), first filial generation (F1), second filial generation (F2). Using the diagrams of pea blossoms in section 12.2, try to determine the genotypes that would produce the phenotypes you see. Use P for the purple (dominant) allele and p for the white (recessive) allele, and remember that each individual has two parents, so has alleles for any one trait (one from each parent). (answers below)
- Define: dominant allele, recessive allele.
- Using Figure 12-15, state why gametes have only ONE copy of each gene. State the possible gametes that can be produced by individuals with these geneotypes: PP, Pp, pp (answer below -- if you don’t get it, study the diagrams in this section).
- What possible offspring could be produced by these pea crosses? Use either Punnett squares or mathematical probabilities to predict both phenotypic (appearance) and genotypic ratios. (answers below)
  Purple PP x white pp
  Purple Pp x white pp
  Purple Pp x purple PP
  Purple Pp x purple Pp
- Define incomplete dominance. Notice that the genotypic ratios are the same as they are for a simple monohybrid cross, but that both alleles show simultaneously in the phenotype of the heterozygous individuals.
- List the traits that Mendel studied in pea plants.
- What ratios of traits did Mendel discover in the F2 offspring when he tracked two traits at once?
- State the Law of Independent Assortment.
- In diagram 12-14, what do the circles with letters (on top of and down the side of the Punnett square) represent? Why is there only one of each of the two alleles in these circles?

Problems

1. Remember Kurt and Shana from the in-class movie? Shana has cystic fibrosis. Her husband, Kurt, was tested for the CFTR gene and found out he is a carrier (heterozygous). Use C for the dominant (normal) form of this gene and c for the recessive (disease) allele, and predict the odds of Kurt and Shana having a child who a) has CF, b) is normal, but a carrier for CF, c) is normal and not a carrier.
2. Huntington's disease is a rare but fatal disease of the nervous system caused by a dominant allele. The symptoms of the disease do not show up until about age 40 or 50, so it can be inherited and passed on before the disease claims its victim. Using H for the disease allele and h for the normal allele, predict the odds of a child inheriting Huntington's disease from a) a mother who is homozygous recessive and a father who is heterozygous, b) a mother who is homozygous recessive and a father who is homozygous dominant.
3. Flower color in snapdragons shows incomplete dominance. A cross between a pure-breeding red flowering plant and a pure-breeding white flowering plant produces all pink flowers. Is it possible to get a pure-breeding pink snapdragons?
4. List the possible gametes that can be produced by each of these individuals:
   Aa BB
   AA bb
   aa Bb
   Aa Bb
5. In guinea pigs, the allele for short hair (S) is dominant over long hair (s), and the allele for black hair (B) is dominant over brown hair (b). Suppose two guinea pigs that are heterozygous for both traits are crossed. What are the odds that any one of their pups will be:
   a. short-haired and black?
   b. short-haired and brown?
   c. long-haired and black?
   d. long-haired and brown?
6. Suppose that a guinea pig boar that is heterozygous for hair length and hair color is bred to a sow that is heterozygous for hair (Ss) but whose hair is brown (bb). What are the odds that any one of their pups will be:
   a. short-haired and black?
   b. short-haired and brown?
   c. long-haired and black?
   d. long-haired and brown?

Useful websites:

- "Interactive monohybrid cross" http://science.nhmccd.edu/biol/monohybr/monhybr.html is an interactive site that teaches how to do monohybrid (single gene) crosses.
- "Incomplete dominant interactive set" http://www.ksu.edu/biology/pob/genetics/incom.htm is an interactive website that teaches about incomplete dominance.
- "Dihybrid Crosses" http://science.nhmccd.edu/biol/dihybrid/dihybrid.html is an interactive tutorial to help you understand how to set up a dihybrid Punnett square.
- "Dihybrid Cross Tutorial" http://www.biology.arizona.edu/mendelian_genetics/problem_sets/dihybrid_cross/03t.html takes you step-by-step through making a dihybrid Punnett square.

Answers:

Reading notes:

Parental generation: purple flowered plants = PP, white-flowered plants = pp

F1 generation: All purple, all Pp

F2 generation: 3/4 purples , 1/4 white. Genotypes: 1/4 PP, 1/2 Pp, 1/4 pp

Gametes: PP --> P gametes only

Pp --> P or p gametes

pp --> p gametes only

Crosses: Purple PP x white pp = All purple; all pp

Purple Pp x white pp = 1/2 purple, 1/2 white; 1/2 Pp, 1/2 pp

Purple Pp x purple PP = All purple; 1/2 PP, 1/2 Pp

Purple Pp x purple Pp = 3/4 purple, 1/4 white; 1/4 PP, 1/2 Pp, 1/4 pp

Mendel discovered the 9:3:3:1 ratio: 9 dominant/dominant, 3 dominant/recessive, 3 recessive/dominant, 1 recessive/recessive.

The circles on the top and sides of the square represent the gametes of the parents. There are two letters in each because we are tracking two independent traits at once.

Problems:

1. CF child (cc)= 1/2; normal but carrier child (Cc) = 1/2; normal, not a carrier (CC) = 0.
2. a. hh x Hh --> 1/2 hh (normal), 1/2 Hh (affected) b. hh X HH --> all Hh, all affected
3. No. Pink-flowered snapdragons are always heterozygous (RR’), and so if they are crossed, will always produce 1/4 red offspring, 1/2 pink, and 1/4 white. The only way to get all pink flowers in a generation is to cross red and white-flowering plants.

4. Possible gametes:
   Aa BB --> AB or aB
   AA bb --> Ab
   aa Bb --> aB or ab
   Aa Bb --> AB or Ab or aB or ab

5. Both parents will have genotypes Ss Bb for these two traits. Both can have these gametes: SB, Sb, sB, sb
   a. short-haired and black = 9/16
   b. short-haired and brown = 3/16
   c. long-haired and black = 3/16
   d. long-haired and brown = 1/16

   The Punnett square looks like this (bold = short/black, underline = short/brown, italic = long, black plain = long, brown)

	SB	Sb	sB	sb
SB	**SSBB**	**SSBb**	**SsBB**	**SsBb**
Sb	**SSBb**	SSbb	**SsBb**	Ssbb
sB	**SsBB**	**SsBb**	*ssBB*	*ssBb*
sb	**SsBb**	Ssbb	*ssBb*	ssbb

6. In this case, the boar's genotypes is SsBb, but the sow's is Ssbb. This will produce a different Punnett square.
   Boar's possible gametes: SB, Sb, sB, sb
   Sow's possible gametes: Sb, sb

   A Punnett square for this cross looks like:

	SB	Sb	sB	sb
Sb	**SSBb**	Ssbb	**SsBb**	Ssbb
sb	**SsBb**	Ssbb	*ssBb*	ssbb

And the resulting ratios for the pups are:
a. short-haired and black = 3/8
b. short-haired and brown = 3/8
c. long-haired and black = 1/8
d. long-haired and brown = 1/8