TwoOldGuys™ Study Guides BI114 Biological Concepts for Teachers Chapter 3. Inheritance and Evolution 3.1. Genetics

Based on Indiana's Academic Standards, Science, as adopted by the Indiana State Board of Education, Nov 2000. Numbers refer to the age-appropriate grade-level for the content.

Review

Before, when our interest was in the species and the higher level taxa, we were concerned with how individual animals were like other members of their species, genus, family, order, etc. It was the similarities among individual members of the species which we used to establish the characteristics of the species of which our individual animal was a member. Likewise, the similarities among the species of the genus, among the genera of the family, among the families of the order, are the characteristics of the higher taxa. We have reviewed the major groups of plants and animals, and a few of the less familiar types.

Now, we are interested in the characteristics below the species level, and with the behavior of the characteristics over rather long time frames, although not the very long Geologic times of the previous chapter. Rather, we will be looking at the characteristics of Living things [organisms] over several generations. At this level, we switch from similarities (compare) to differences (contrast).

Genetics

Inheritable characteristics

grade K: to 3:

There may be differences among individuals of the same type (species) of animal

For the youngest of your students, we return to the familiar pet-type animals. The children will most likely have seen enough dogs or cats to realize that not all of them are alike. Dogs, for example, differ in size, color, hair length, markings.... More subtle differences include head shape, tail length, relative size of shoulders compared to hips, etc. If you ask them about cats, your students should be aware of differences in hair length, markings, and weight among other characteristics. However, should you ask your students to describe specific characteristics of robins that differ from one individual to another, you should not be surprised to get no good answers. It is not that robins are less different from each other; after all, a female robin can identify her mate, and will chase off other males. The problem with humans' ability to identify individual robins is mostly a matter of interest; most of us simply don't care which robin is which.

grade 3: to 4:

Recognize that litters are like parents and like brothers & sisters

Those differences which help us identify individual animals tend to run in families, not in the taxonomic sense, but in the sense of parents and children. A litter of puppies (say beagles) look more like the parents than like a litter from different parents (say retrievers). Things get more interesting (remember, we defined 'interesting' to mean anything that I find interesting) when we look at litters of mutts [non-pedigreed dogs]. In such a litter, some pups will look more like one parent while other pups look like the other parent. Some may even have some characteristics from each parent. In this case, the pups don't match either parent but combine characteristics of both.

grade 5: to 6:

Recognize that there are patterns in inheritance

By this age group, the students will be able to understand the patterns in inheritance, and should be able to develop tentative explanations of the patterns. The key features to be noted are that pure bred (pedigreed) dogs produce similar puppies that resemble the parents. The parents are able to pass information to the children to make them resemble the breed standard. On the other hand, mixed-breed dogs produce dissimilar puppies. Some of these puppies will resemble one parent; others, the other parent; while other puppies seem to have mixed traits from both parents. Again the parents appear to pass information about appearance to the puppies, but the puppies choose between the information from the father and that from the mother to determine what to look like.

Information is transmitted from parental generation to offspring generation

According to the Indiana State Standards, the correct way to present this is as follows:

- both sexes in the parental generation (P-1) make specialized cells to carry that information to the offspring
- the fertilized egg combines the information from the P-1 to form the offspring generation (F-1).

[This may be viewed as "sex education" by some parents.]

An alternative that may avoid the 'sex education' stigma is to ignore the 'specialized cells.' The parents (P-1) still pass information in some physical form to the offspring (F-1) and this information is used to establish the characteristics of the offspring individuals. The offspring receive information equally from each parent, who in turn received the information from both of their parents. Sometimes the information from one parent is stronger than that from the other parent, so the offspring look a lot like the parent with the stronger information. However the weaker information is still there, so the trait can "skip a generation," resulting in children which resemble their grandparents.

The story is basically the same without or with "sex." Information is passed from generation to generation in some physical form or in the form of specialized [sex] cells.

Some characteristics are acquired, not inherited

Some traits are "clearly" inherited, such as human eye color. The color of a human's eyes is determined from the information transmitted from the parents, thus inherited. Some characteristics are "clearly"

acquired, such as extent of muscle development in humans. The amount of muscle development depends almost entirely by the amount of time spent in strenuous physical activity, such as lifting weights. For many human characteristics, there has been a long standing argument between "nature" versus "nurture," or inherited [nature] versus acquired [nurture]. The two complications driving these arguments are first, that many inherited traits are influenced by environment, and second, that many acquired characteristics are limited by inheritance. As an example of the first, we think that basic intelligence is inherited, but well educated parents provide greater education than do poorly educated parents, so the amount of inherent intelligence is difficult to separate from amount of education. As an example of the second, we think that the propensity to gain excess weight is inherited, so some people gain weight easily and lose it with difficulty, but diet and exercise are far more important in determining adult body weight.

grade 7: to 8: and secondary: to college:

Mendel's Principles

Gregor Mendel was a Monk whose duties at the monastery included growing the vegetables to feed the Brothers [other Monks at the monastery]. He understood that the growing of vegetables also meant saving enough seed for next years crop. It is apparent that he also read documents on the principles of agricultural breeding. Incidentally, the oldest known such document was found at an archeological site in Mesopotamia (currently northern Turkey, near the Black Sea) in a stratum dated to about 5,000 BP. This document is a "how-to" manual, basically 'animal breeding for dummies.' It seems reasonable to assume that we had to have considerable experience in breeding before we attempted to write the 'How-To' manual. Prior to Mendel, the best

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theoretical version of inheritance was very similar to the description above under grades 5 to 6. Because Mendel had lots of time to think about inheritance, and because Mendel was unusually brilliant, he was able to come up with the only true theory (model) we have in Biology, although he described it (in translation from the Austrian) as "Principles of Inheritance." Mendel's Theory of Inheritance consists of the following Principles, rewritten using the vocabulary from section 3.0:

- 1. Each individual receives one allele for each trait from each parent.
- 2. [therefore] each individual has two alleles for each trait.
- 3. alleles are segregated during gamete formation.
- 4. alleles are combined during fertilization.
- 5. one allele (dominant) may mask the expression of the other (recessive).

Notice that principle 1 is a hypothesis. Principle 2 is actually a logical conclusion deduced from the hypothesis (principle 1). Principles 3 and 4 are best considered to be predicted observable events which ought to occur if the hypothesis is true. Principle 5 is another hypothesis.

The method most commonly taught for evaluating Mendelian breeding experiments is Punnett's method.

The Cute & Fuzzy Bunnies[™] are borrowed from the movie *One Crazy Summer* © 1986 Warner Bros, in which the Cute & Fuzzy Bunnies[™] appear as cartoon characters. For our purposes, Bugs Bunny [another Warner Bros cartoon cwazy wabbit] is considered to be one of the Cute & Fuzzy Bunnies[™] species. These imaginary creatures are introduced here to illustrate the principles of inheritance [the science of Genetics]. The Cute & Fuzzy Bunnies[™] have an advantage over real creatures for introductory Genetics because I can make up whatever traits I wish simply because no one can show any breeding data to contradict my assumption(s) because the Cute & Fuzzy Bunnies[™] are cartoon creatures. Equally important, the traits of imaginary creatures behave according to the 'rules.' Human genetics, by contrast, tend to ignore the rules, a concept which is explained at the end of this section.

The simplest traits of the Cute & Fuzzy Bunnies[™] do not exhibit dominance, so all three genotypes can be identified from the phenotype. For example, if the interesting trait is height:

TT = tall (Bugs Bunny is an example)
Tt = medium height (chin-height compared to Bugs Bunny)
tt = short (able to stand upright under Bugs Bunny's outstretched
arm).

Here the trait is height, and the expressions are tall, medium and short. The alleles are T for tallness, and *t* for shortness. The tall expression results from a 'double' dose of tallness, TT; while the short expression results from a complete absence of tallness, *tt*. A single dose of tallness produces a slightly tall (or medium) individual, T*t*.

Mendelian dominance occurs when the heterozygous individual has the same phenotype as one of the two homozygous individuals. For example in the Cute & Fuzzy Bunnies[™], shaggy hair (long hair) is dominant over short hair:

HH = shaggy hair H*h* = shaggy hair *hh* = short hair

Here the interesting trait is hair length, but there are only two expressions: shaggy (long) and short. Because of dominance, any allele(s) for longness, H, produces long hair; while again the absence of longness, *hh*, results in short hair.

The following series of tables illustrates the analysis of Mendel's predictions using Punnett's method.

Punnett Analysis of monohybrid

1. List the parents (P-1) by phenotype and by genotype, separated by an X, in the box on the left:

medium male X medium female								
Tt	Х	Tt						
				summary				
				TT	_	tall	_%	
				Tt	_	medium	_%	
				tt		short	_%	
				totals	_		100%	

2. Record the gametes::

a. the mother's gametes are column labels for the Punnett Square, and are used as column entries:

medium male X medium female								
Tt	Х	Tt						
				summary				
	Т	t		TT		tall	_%	
	T_	_t		Tt	_	medium	_%	
	T_	_t		tt		short	_%	
				totals			100%	

b. the father's gametes are the row labels for the Punnett Square, and are used as the row entries, completing the zygotes:

medium male X medium female								
Tt	Х	Tt						
				summary				
	Т	t		TT		tall	_%	
Т	TT	Tt		Tt		medium	_%	
t	Tt	tt		tt		short	_%	
				totals			100%	

3. Tally the resulting genotypes [TT, Tt, tt] for the offspring (F-1):

4. Tabulate the resulting phenotypes [tall, medium, short] with percentages:

medium male X medium female								
Tt	Х	Tt						
				summary				
	Т	t		TT	1	tall	25%	
Т	TT	Tt		Tt	2	medium	50%	
t	Tt	tt		tt	1	short	25%	
				totals	4		100%	

Monohybrid with Dominance

Mendelian dominance occurs when the heterozygous individual has the same phenotype as one of the two homozygous individuals. For example in the Cute & Fuzzy Bunnies[™], shaggy hair (long hair) is dominant over short hair. We symbolize this trait as H for long hair, h for short hair.

hybrid shaggy father X hybrid shaggy mother

hybrid 1	hybrid male X hybrid female							
Hh	Х	Hh						
				summary				
				HH		Shaggy	_%	
_				Hh				
				hh	_	short	_%	
				totals			100%	

hybrid male X hybrid female							
Hh	Х	Hh					
				summary	7		
	Η	h		HH	3	Shaggy	75%
Η	HH	Hh		Hh			
h	Hh	hh		hh	1	short	25%
				totals			100%

Patterns of Inheritance predicted by Mendelian principles

For traits with no dominance, all three genotypes can be recognized because each has its own phenotype. In this case a pure-bred cross, such as pure-bred tall crossed to pure-bred tall [TT X TT], or such as pure-bred short crossed to pure-bred short [tt X tt], produces Cute & Fuzzy BunniesTM offspring that closely resemble their parents in terms of height; tall from tall X tall, short from short X short. The hybrid cross, medium crossed to medium [Tt X Tt] produces a mixed litter with tall, medium and short Cute & Fuzzy BunniesTM. This pattern was described above as simple, because this is the full story. The parental [P-1] generation mating could be

P-	-1	F-1			
male	female	TT	Tt	tt	
tall (TT)	tall (TT)	100% tall	0 med	0 short	
tall (TT)	med (T <i>t</i>)	50% tall	50% med	0 short	
tall (TT)	short (<i>tt</i>)	0 tall	100% med	0 short	
med (Tt)	tall (TT)	50% tall	50% med	0 short	
med (T <i>t</i>)	med (T <i>t</i>)	25% tall	50% med	25 % short	
med (T <i>t</i>)	short (<i>tt</i>)	0 tall	50% med	50% short	
short (<i>tt</i>)	tall (TT)	0 tall	100% med	0 short	
short (<i>tt</i>)	med (Tt)	0 tall	50% med	50% short	
short (<i>tt</i>)	short (<i>tt</i>)	0 tall	0 med	100% short	

Where it becomes simple is that we can substitute the offspring generation (F-1) for the parental generation (P-1), and the second offspring generation (F-2) for the first offspring generation (F-1), and the entire table does not change.

Dominance adds considerable complication to the analysis: shaggy could be either HH or Hh, but short-hair can be only hh. As a result,

• shaggy X shaggy may produce all shaggy

- shaggy X shaggy may produce 75% shaggy/25% short-hair
- shaggy X short-hair may produce all shaggy
- shaggy X short-hair may produce 50% shaggy/50% short-hair.
- short-hair X short hair always produces short-hair.

In summary, most matings involving dominance have multiple possible outcomes.

P-1	F-1	F-2
shag X shag	shag	shag
	shag	shag/short
	shag/short	shag/short
shag X short	shag	shag/short
	shag/short	shag/short
short X shag	shag	shag/short
	shag/short	shag/short
short X short	short	short

You should note in this table that short-hair is true breeding; short X short always produces short-hair. Sometimes the short-hair expression skips a generation in a shaggy X short (or short X shaggy) cross, but not always. Even in a shaggy X shaggy cross, short-hair may appear "out of nowhere." When short-hair appears out of nowhere, it means that there are short-hair Cute & Fuzzy Bunnies[™] somewhere in the family tree.

The following table shows the genotypes of the parental (F-1) generation, and the possible genotypes for the F-1 generation. To determine the possible genotypes for the F-2 generation you must consider all possible matings among the F-1 generation. You should be able to do the analysis necessary to confirm the possible F-2 offspring described in the table.

P-1	F-1	F-1 matings	F-2
shag X shag	shag	HH x HH	shag
HH X HH	4 HH		
shag X shag	shag	HH x HH	shag/short
HH X Hh	2 HH, 2 Hh	HH x Hh	
		Hh x Hh	
shag X shag	shag/short	HH x HH	shag/short
Hh X Hh	1 HH, 2 Hh/1 hh	HH x Hh	
		hh x hh	
shag X short	shag	Hh x Hh	shag/short
HH X hh	4 Hh		
shag X short	shag/short	Hh x Hh	shag/short
Hh X hh	2 Hh/2 hh	Hh x hh	
		hh x hh	
short X shag	shag	Hh x Hh	shag/short
hh X HH	4 Hh		
short X shag	shag/short	Hh x Hh	shag/short
hh X Hh	2 Hh/2 hh	Hh x hh	
		hh x hh	
short X short	short	hh x hh	short
hh X hh	4 hh		

Human Genetics

You need to remember that in section 1.1 "The Science Process" it was suggested that science functions in large part by asking only simple questions, the answers to which lead to somewhat more complex questions. It was also defined that theories are also known as models, suggesting that they [as models] resemble only the most important aspects of the reality they represent, thereby leaving out some of the details. Science often advances by incorporating more details into our models. These two concepts can explain why Mendelian Genetics do not describe Human Genetics very well.

Recall that most people have difficulty telling one robin from another, yet can easily recognize their pet dog. The reason is that most people have no interest in determining *which* robin they are looking at, so look for characteristics which identify the beast as a robin [assuming, perhaps inaccurately, that they even care what *kind* of bird it is]. These same people care far more for their pets, and look for traits which distinguish their pet from other pets of the same kind. We have sufficient interest in our own species that we seek even trivial details to distinguish individuals [sometimes including identical twins].

Mendel's Principles are the answer to the simplest question concerning inheritance: what mechanism allows expressions of some traits to be passed from one generation (P-1) to the next (F-1 or F-2)? Mendel's theory is a model, and was designed to identify a mechanism which accounts for the transmission of genetic information across generations. The principles work rather well for organisms, such as garden peas, where we have limited knowledge of the details of the traits. As our knowledge increases, such as in Humans, the model is too simple to account for our observations.

As an example, consider the traits governing eye color. The brown expression is dominant over blue; blue is dominant over hazel [greygreen-blue]; and hazel is dominant over albino [colorless, appears pink]. Almost every time I have described this to a class of more than fifteen students, at least one is aware of a family where a blue eyed father and hazel eyed mother had a brown eyed child. The brown-blue-hazel-albino color series is composed of three different traits, as follows:

expression	brown(W)/ ~brown(w) ¹	blue(U)/ ~blue(u)	~albino(A)/ albino(a)			
brown	WW, Ww	UU, Uu	AA, Aa			
blue	ww	UU, Uu	AA, Aa			
hazel	WW, Ww, ww	uu	AA, Aa			
albino	WW, Ww,	UU, Uu,	aa			
	ww uu					
¹ tilde (~) means not, as ~Word means "not-Word"						

The blue eyed father would be (ww UU AA) or (ww Uu AA); and the hazel eyed mother would be (WW uu AA) or (Ww uu AA).

- for the brown/~brown trait,
 - o P-1: ww X Ww
 - o F-1: 50% Ww, 50% ww
- for the blue/~blue trait,
 - o P-1: Uu X uu
 - o F-1: 50% Uu, 50% uu
- for the ~albino/albino trait,
 - o P-1: AA X AA
 - F-1: AA
- combining these, the F-1 could be

Ww Uu AA	brown	25%	25%
ww Uu AA	blue	25%	25%
Ww uu AA	hazel	25%	50%
ww uu AA	hazel	25%	

This excessively complex explanation does not account for green eyes, which are yet another trait probably unrelated to the brown-blue series, but does require not-albino [hazel].