Patterns of Inheritance

- In this section we will examine how studying patterns of inheritance has allowed scientists to learn about some of the basic laws that govern the transmission of genetic information from one generation to the next.
- You will also learn how to use this knowledge to predict the outcome of certain types of genetics crosses.

1

Objective # 1

Define the term "monohybrid cross". Describe one of the monohybrid crosses carried out by Mendel and explain how the results of these crosses led him to formulate the Law of Segregation and the Law of Dominance.

2

Objective 1

- So far, we have focused on how chromosomes get passed from cell to cell during the eukaryotic cell cycle, and from one generation to the next during eukaryotic life cycles.
- Since chromosomes contain the hereditary information, this shows us how the hereditary information is transmitted from one generation to the next.

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Objective 1

- However, over 50 years before scientists understood the role that chromosomes play in transmitting hereditary information, an Austrian monk named Gregor Mendel discovered the basic principles of heredity by studying the pattern of inheritance for 7 different traits in pea plants.

4

Objective 1

Each trait that Mendel studied existed in 2 discrete forms. For example, seed shape could be wrinkled or round:

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Objective 1

The 7 traits that Mendel studied are shown in the chart on the right. Each trait exists in 2 forms.

Character	Dominant vs. recessive trait
Flower color	Purple x White
Seed color	Yellow x Green
Seed shape	Round x Wrinkled
Pod color	Green x Yellow
Pod shape	Inflated x Constricted
Flower position	Axial x Terminal
Plant height	Tall x Dwarf

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Objective 1

- Mendel was successful where many before him had failed because:
  - he simplified the problem by first examining just one trait at a time.
  - he limited his study to traits that existed in 2 discrete forms.
  - he kept quantitative data over several generations, using large sample sizes.

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Objective 1

- he used statistics to mathematically analyze the results of his crosses and help determine the general patterns of his results.

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Objective 1

- Mendel's success was also due to a certain amount of luck because each trait that he followed happened to be controlled by a single pair of alleles. This produces the simplest possible pattern of inheritance.

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Objective 1

- Mendel began with crosses where he followed the inheritance of one trait. Although he didn't know it, each trait he studied was controlled by a single gene locus. This type of cross is called a monohybrid cross.
- For example, Mendel followed the inheritance of flower color, which exists in 2 discrete forms: purple and white.

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Objective 1

- When he crossed some pure-breeding purple flowered plants with some pure-breeding white flowered plants (this is called the parental or P generation) he got some surprising results:
  - all of the offspring (called the F_1 generation) had purple flowers!

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Objective 1

- Next, he allowed the F_1 plants to self-fertilize in order to produce the F_2 generation. Again, the results were unexpected:
  - Out of 929 F_2 plants, he found 705 with purple flowers and 224 with white flowers, a ratio of 3.15 to 1.

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Objective 1, One of Mendel's Monohybrid Crosses

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Objective 1

Mendel explained his results by proposing the following 4-part hypothesis:

1) Each individual has two "hereditary factors" controlling a given trait. The pure-breeding purple parents have 2 hereditary factors for purple flowers, and the pure-breeding white plants have 2 hereditary factors for white flowers.

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Objective 1

- Today we call Mendel's "hereditary factors" alleles. Scientists use letters to represent alleles. For example, if we use "A" to represent the purple allele and "a" to represent the white allele, then the pure-breeding purple plants would be AA and the pure-breeding white plants would be aa.

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Objective 1

- The appearance of an organism is called its phenotype (e.g. purple flowers), and the alleles the organism has is its genotype (e.g. AA).

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Objective 1

- From chromosome studies we know that parents are diploid, so there are 2 sets of chromosomes in each cell, and 2 alleles at each gene locus.
- Because flower color is controlled by one gene locus, each parent must have 2 alleles controlling this trait.

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Objective 1

- If the 2 alleles are identical (e.g. AA), the individual is homozygous for that trait. If the 2 alleles are different (e.g. Aa), the individual is heterozygous for that trait.

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Objective 1

2) When the parents produce gametes, the 2 hereditary factors separate, and each gamete receives one of the 2 factors. Therefore, all gametes produced by the purple parent (AA) have one purple allele (A), and all gametes produced by the white parent (aa) have 1 white allele (a). This is called Mendel's Law of Segregation.

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Objective 1

- Today we know that homologous chromosomes separate during meiosis I, leading to formation of haploid gametes.
- Because gametes are haploid, each gamete has 1 set of chromosomes, and 1 allele at every gene locus.
- Because flower color is controlled by one gene locus, each gamete must have 1 allele controlling this trait.

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Objective 1

3) The offspring are formed when a gamete from one parent joins with a gamete from the other parent. Therefore, each F_1 offspring receives one purple hereditary factor (A) from the purple parent (AA) and one white hereditary factor (a) from the white parent (aa).

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Objective 1

- Today we know the offspring are diploid, so there are 2 sets of chromosomes in each cell, and 2 alleles at each gene locus (one inherited from each parent).

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Objective 1

4) When an individual is heterozygous, only one of the 2 alleles is expressed. Mendel called the expressed allele dominant, and the non-expressed allele recessive. Because purple is dominant to white, all the F_1 plants (Aa) have purple flowers. This is known as Mendel's Law of dominance.

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Objective 1

- One the next slide, you can see how Mendel's Laws of Inheritance can be used to correctly predict the outcome of both the F_1 and the F_2 generations:

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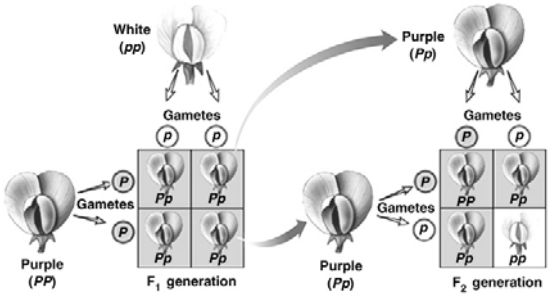
Objective 1, One of Mendel's Monohybrid crosses



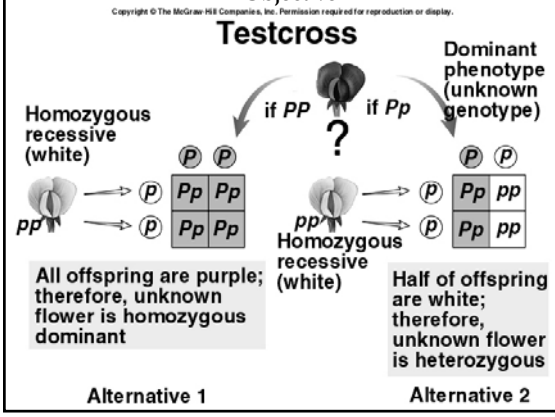
Objective # 2

Explain how a testcross can be used to determine whether an individual with a dominant phenotype is homozygous or heterozygous.

Objective 2

- To determine whether an individual with a dominant phenotype is homozygous for the dominant allele or heterozygous, Mendel crossed the individual in question with an individual that had the recessive phenotype:

Objective 2



Objective # 3

Define the term "dihybrid cross". Describe one of the dihybrid crosses carried out by Mendel and explain how the results of these crosses led him to formulate the Law of Independent Assortment.

Objective 3

- In a dihybrid cross, we follow alleles at 2 gene loci simultaneously:
  - the parents are diploid, so there are 2 alleles at each gene locus = 4 alleles total
  - the gametes are haploid, so there is 1 allele at each gene locus = 2 alleles total
  - the offspring are diploid, so there are 2 alleles at each gene locus = 4 alleles total

Objective 3

- For example, in pea plants seed shape is controlled by one gene locus where round (R) is dominant to wrinkled (r) while seed color is controlled by a different gene locus where yellow (Y) is dominant to green (y).
- Mendel crossed 2 pure-breeding plants: one with round yellow seeds and the other with green wrinkled seeds.

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Objective 3

- With his monohybrid crosses, Mendel determined that the 2 alleles at a single gene locus segregate when the gametes are formed.
- With his dihybrid crosses, Mendel was interested in determining whether alleles at 2 different gene loci segregate dependently or independently.

32

Objective 3

- Dependent segregation (assortment) means alleles at the 2 gene loci segregate together, and are transmitted as a unit. Therefore, each plant would only produce gametes with the same combinations of alleles present in the gametes inherited from its parents:

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Objective 3, Dependent Assortment

Parents → R R / Y Y r r / y y

Parental Gametes → R / Y r / y

F_1 Offspring → R r / Y y

F_1 Offspring's Gametes → R / Y r / y

What is the expected phenotypic ratio for the F_2?

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Objective 3

F_2 with dependent assortment:

	R / Y	r / y
R / Y	R R / Y Y	R r / Y y
r / y	R r / Y y	r r / y y

Ratio is 3 round, yellow : 1 wrinkled, green

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Objective 3

- Independent segregation (assortment) means alleles at the 2 gene loci segregate independently, and are NOT transmitted as a unit. Therefore, each plant would produce some gametes with allele combinations that were not present in the gametes inherited from its parents:

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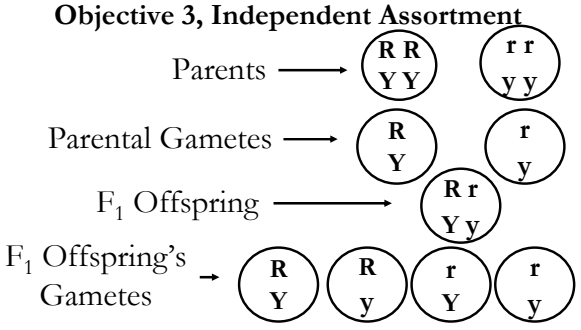
Objective 3, Independent Assortment



What is the expected phenotypic ratio for the F_2?

Objective 3

F_2 with independent assortment:

	RY	Ry	rY	ry
RY	RR YY	RR Yy	Rr YY	Rr Yy
Ry	RR Yy	RR Yy	Rr Yy	Rr yy
rY	Rr YY	Rr Yy	rr YY	rr Yy
ry	Rr Yy	Rr yy	rr Yy	rr yy

Phenotypic ratio is 9 : 3 : 3 : 1

Objective 3

- Mendel's dihybrid crosses showed a 9:3:3:1 phenotypic ratio for the F_2 generation.
- Based on these data, he proposed the Law of Independent Assortment, which states that when gametes form, each pair of hereditary factors (alleles) segregates independently of the other pairs.

Objective # 4

Explain why some genes do NOT assort independently. Also explain how an experiment by Morgan originally demonstrated this.

Objective 4

- In the early 20th century, Thomas Hunt Morgan studied patterns of inheritance for many traits in the fruit fly, *Drosophila*.
- At one gene locus controlling eye color, he found a dominant allele for red eyes (R) and a recessive allele for purple eyes (r).

Objective 4

- At another gene locus controlling wing length, he found a dominant allele for normal wings (N) and a recessive allele for short wings (n).
- Morgan crossed a female heterozygous at both gene loci with a male homozygous for both recessive alleles. Based on Mendel's hypotheses, what is the expected outcome?

Objective 4

- Genotype of parents:
  - ♀ Rr Nn X ♂ rr nn
- Genotype of gametes:
  - ♀ (RN) (Rn) (rN) (rn) X ♂ (rn)
- Genotype of offspring:

	RN	Rn	rN	rn
rn	RrNn	Rrnn	rrNn	rrnn

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Objective 4

- Offspring:

genotype	RrNn	Rrnn	rrNn	rrnn
phenotype	red, normal	red, short	purple, normal	purple, short
expected	25%	25%	25%	25%
observed	47%	5%	5%	43%

- Why don't the observed results agree with the predicted results?
- Chance or Mendel's hypothesis is wrong.

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Objective 4

- A statistical test gives a $p < 0.01$. What should we conclude?
  - There is a very small probability the difference was caused by chance alone.
- So, what's wrong with the hypothesis?

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Objective 4

	RN	Rn	rN	rn
rn	RrNn	Rrnn	rrNn	rrnn
expected	25%	25%	25%	25%
observed	47%	5%	5%	43%

- Female gametes are mostly RN or rn. Why didn't we get 25% of RN, Rn, rN, and rn?
- R and N are linked together on one chromosome, while r and n are linked together on the homologous chromosome.

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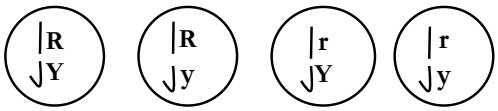
Objective 4

- Unlinked genes (located on different chromosomes) assort independently:

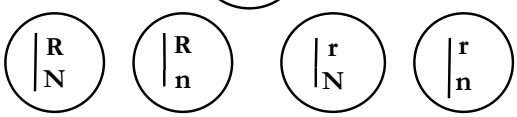


25% 25% 25% 25%

47

Objective 4

- Linked genes (located on the same chromosome) do not assort independently:



47% 5% 5% 43%

48

Objective # 5

Explain how the process of crossing over leads to recombinant types. Explain how the frequency of recombinant types is used by geneticists to construct chromosome maps.

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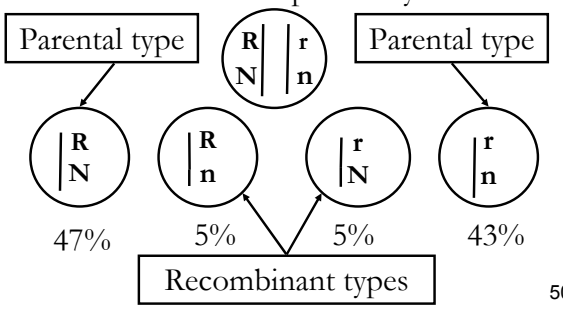
Objective 5

- Linked genes (on the same chromosome) do NOT assort independently:



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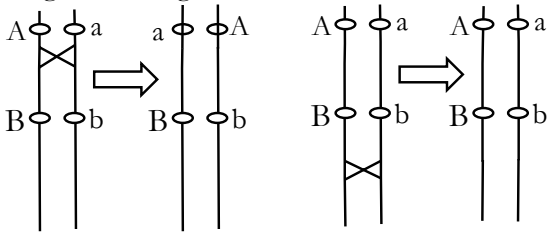
Objective 5

- Recombinant types are produced only when a crossover occurs between the 2 genes being studied:



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Objective 5

- Because crossovers occur along the length of a chromosome at random, the closer 2 genes are, the smaller the chance a crossover will occur between them, and the lower the frequency of recombinant types.
- Therefore, the frequency of recombination can be used to construct genetic maps.

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Objective 5

- One map unit is defined as the distance between 2 genes that will produce 1% recombinant types.
- What is the distance in map units between the 2 gene loci Morgan studied:

RrNn	Rrnn	rrNn	rrnn
47%	5%	5%	43%

➢10 map units

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Objective # 6

Explain how each of the following patterns of inheritance represents a modification of Mendel's original principles:

a) sex linkage
b) incomplete dominance
c) codominance
d) multiple alleles
e) polygenic traits
f) epistasis
g) pleiotropy
h) environmental effects on gene expression

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Objective 6a

a) Sex linkage:

- Sex in humans (and many other organisms, but not all organisms) is determined by genes located on a pair of chromosomes called the sex chromosomes.
- all other chromosomes are called autosomes.

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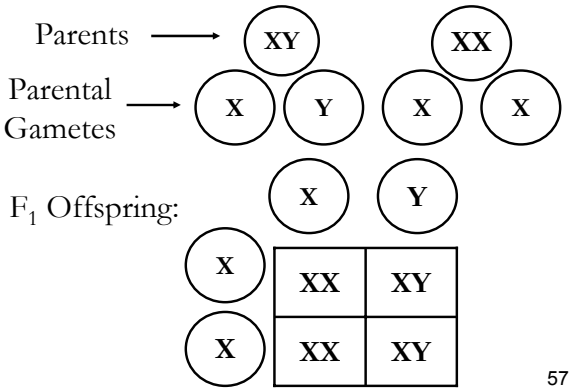
Objective 6a

- Even though the sex chromosomes pair during synapsis, they are not homologous. The larger chromosome is called the X and the smaller is the Y.
- In humans, XX is female and XY is male.
- What is the expected sex ratio among the offspring when we cross a male and female?

56

Objective 6a



57

Objective 6a

- Other traits, besides sex, are controlled by genes on the sex chromosomes. These are called sex-linked traits.
- Traits controlled by the X are X-linked.
- Traits controlled by the Y are Y-linked.
- Since most sex-linked traits are controlled by the X, you can assume X-linkage, unless it says Y-linked.

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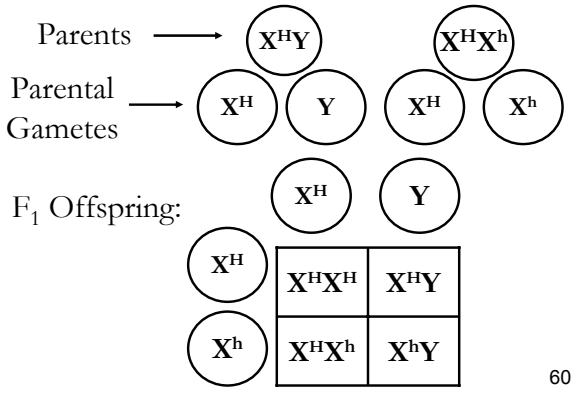
Objective 6a

- X-linked traits are an exception to Mendel's laws because females have 2 alleles for each X-linked trait, but males have only 1.
- In humans, hemophilia is caused by a recessive allele on the X chromosome. Two normal parents have a son with hemophilia. What is the probability their next child will also have hemophilia?

59

Objective 6a



60

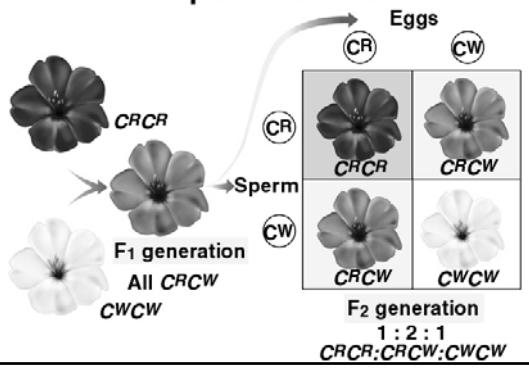
Objective 6b

b) Incomplete dominance:

- neither allele is dominant and heterozygous individuals have an intermediate phenotype
- for example, in Japanese four o'clock, plants with one red allele and one white allele have pink flowers:

61

Objective 6b



62

Objective 6c

c) Codominance:

- neither allele is dominant and both alleles are expressed in heterozygous individuals
- we will examine an example of codominance when we discuss human ABO blood types

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Objective 6d

d) Multiple alleles:

- when there are more than 2 possible alleles at a given gene locus (even though each diploid individual has only 2).
- the human gene locus that controls ABO blood type involves multiple alleles and codominance.

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Objective 6d

- This gene (designated I) codes for an enzyme that adds sugar molecules to lipids on the surface of red blood cells.
- There are 3 possible alleles at this gene locus:

  I^A adds galactosamine

  I^B adds galactose

  i adds neither sugar

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Objective 6d

- These sugars act as recognition markers (antigens) for the immune system.
- The immune system will produce antibodies against cells with foreign antigens and mark them for destruction.

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Objective 6d, ABO Blood Type

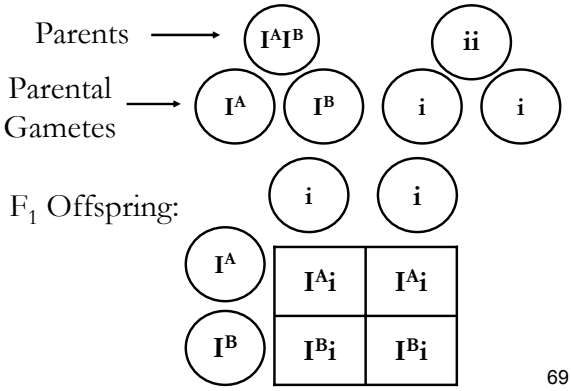
Antigens present	Blood Type	Possible genotypes
galactosamine only	A	$I^A I^A$ $I^A i$
galactose only	B	$I^B I^B$ $I^B i$
both	AB	$I^A I^B$
neither	O	ii

I^A and I^B are codominant

Objective 6d

- If a person with type AB blood marries a person with type O, what blood types are possible among the offspring?

Objective 6d

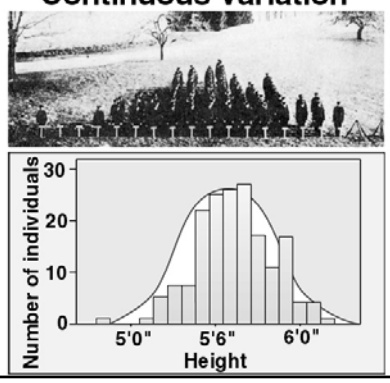


Objective 6e

e) Polygenic traits:

- most traits are not controlled by a single gene locus, but by the combined interaction of many gene loci. These are called polygenic traits.
- polygenic traits often show continuous variation, rather then a few discrete forms:

Objective 6e



Objective 6f

f) Epistasis:

- this is a type of polygenic inheritance where the alleles at one gene locus can hide or prevent the expression of alleles at a second gene locus.
- for example, in Labrador retrievers one gene locus affect coat color by controlling how densely the pigment eumelanin is deposited in the fur.

Objective 6f

- a dominant allele (B) produces a black coat while the recessive allele (b) produces a brown coat
- however, a second gene locus controls whether any eumelanin at all is deposited in the fur. Dogs that are homozygous recessive at this locus (ee) will have yellow fur no matter which alleles are at the first locus:

Objective 6f

Epistatic Interactions on Coat Color

ee	*E_*		
No dark pigment in fur	Dark pigment in fur		
Yellow Lab			
eebb	*eeB_*	*E_bb*	*E_B_*
		Chocolate Lab	***Black Lab***
Yellow fur, brown nose, lips, eye rims	Yellow fur, black nose, lips, eye rims	Brown fur, nose, lips, eye rims	Black fur, nose, lips, eye rims

Objective 6g

g) Pleiotropy:

- this is when a single gene locus affects more than one trait.
- for example, in Labrador retrievers the gene locus that controls how dark the pigment in the hair will be also affects the color of the nose, lips, and eye rims.

Objective 6h

h) Environmental effects on gene expression:

- the phenotype of an organism depends not only on which genes it has (genotype), but also on the environment under which it develops.

Objective 6h

Environmental Effects on an Allele

Arctic Fox in Winter

Arctic Fox in Summer

Objective 6h

- Although scientists agree that phenotype depends on a complex interaction between genotype and environment, there is a lot of debate and controversy about the relative importance of these 2 factors, particularly for complex human traits.