Module 3C – Patterns of Inheritance

- In this module, we will examine how studying the appearance of traits over several generations has allowed scientists to discover the basic laws that govern the transmission of genetic information from one generation to the next.

1

Objective # 16

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 16

- So far, we have focused on how chromosomes get passed from cell to cell during cell division, and from one generation to the next during eukaryotic life cycles.
- Since chromosomes contain the genetic information, the study of chromosomes provides a key to understanding how the genetic information is transmitted from one generation to the next.

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Objective 16

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

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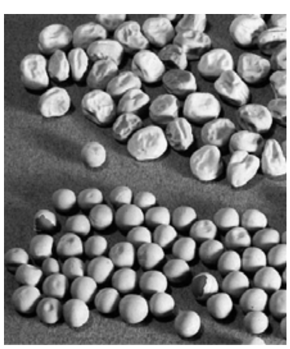
Objective 16

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



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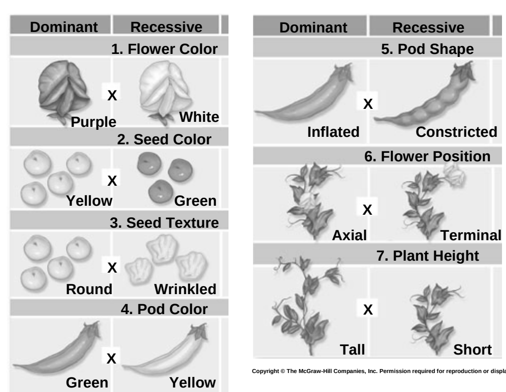
Mendel's Seven Traits



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Objective 16

- 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, and used large sample sizes.

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Objective 16

- 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 16

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

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Objective 16

- 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 one gene locus (a single pair of alleles.) 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 16

- 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 16

- 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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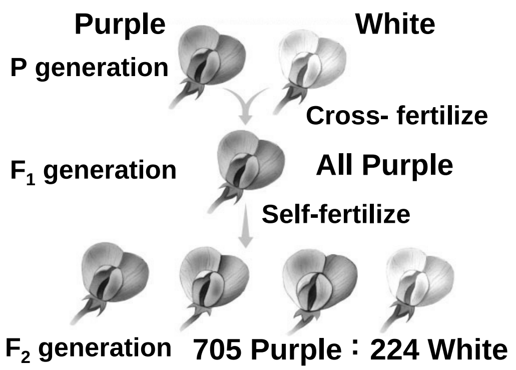
One of Mendel's Monohybrid Crosses



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Objective 16

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 (AA), and the pure-breeding white plants have 2 hereditary factors for white flowers (aa).

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Objective 16

- Today we call Mendel's "hereditary factors" alleles.
- The appearance of an organism is called its phenotype (e.g. purple flowers), and the alleles the organism or gamete has is its genotype (e.g. AA).

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Objective 16

- From chromosome studies we know that the 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 16

- If the 2 alleles are identical, the individual is homozygous for that trait, and if the 2 alleles are different, the individual is heterozygous for that trait:

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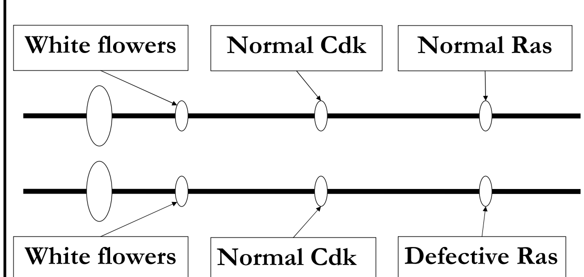
Objective 16

Homologous Chromosomes



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Objective 16

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 16

- 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 16

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 16

- 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 16

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 16

- Follow the procedure on the class handout, "Predicting the Outcome of Monohybrid and Dihybrid Crosses", to determine if Mendel's 4-part hypothesis can accurately predict the outcome of the F_2 generation.

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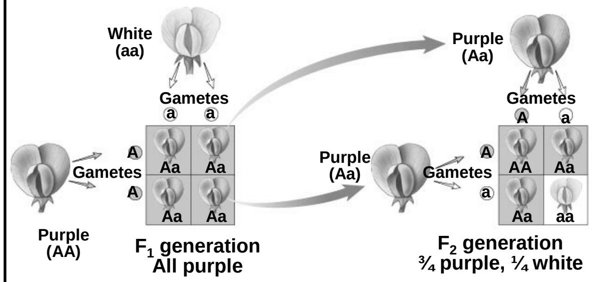
One of Mendel's Monohybrid Crosses



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Objective # 17

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

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Objective 17

- 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:

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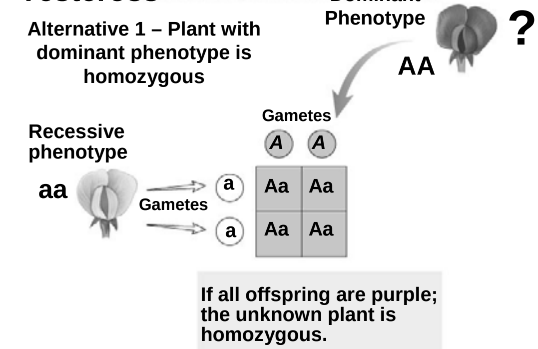
Testcross



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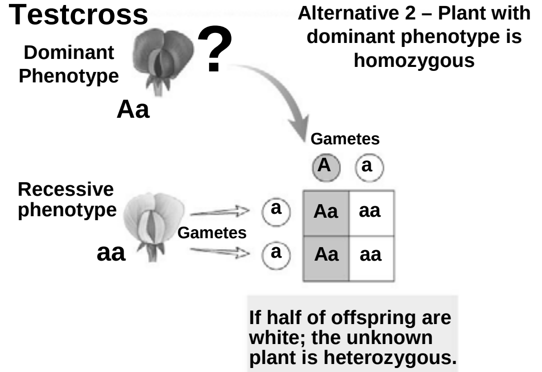
Testcross



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Objective # 18

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.

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Objective 18

- Based on his monohybrid crosses with pea plants, Mendel proposed the Law of Segregation and the Law of Dominance.
- Next, he conducted a series of dihybrid crosses.
- A dihybrid cross is a cross where we follow the inheritance of alleles at 2 different gene loci simultaneously.

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Objective 18

- In a dihybrid cross:
  - 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

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Objective 18

- 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 18

- In the F1 generation, Mendel found that all the offspring had round, yellow seeds.
- In the F2 generation, the approximate proportions were as follows:

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Objective 18

- In order to explain his results, Mendel proposed the Law of Independent Assortment.
- Independent Assortment means alleles at the 2 gene loci segregate independently of each other and are NOT transmitted as a unit. Therefore, each plant can produce gametes with allele combinations that were not present in the gametes inherited from its parents:

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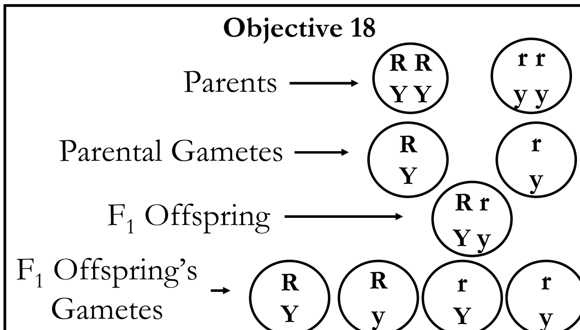
Objective 18



If the allele pairs assort independently, all 4 types of gametes are equally likely.

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Objective 18

F_2 with independent assortment:

Genotypes of gametes produced by F1 generation

	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

Expected phenotypic ratio is 9 : 3 : 3 : 1

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Objective 18

- All of Mendel's dihybrid crosses showed an approximate 9:3:3:1 phenotypic ratio for the F_2 generation.
- Therefore, his results are in agreement with the prediction made based on the Law of Independent Assortment.

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Objective # 19

Explain how each of the following patterns of inheritance represents an exception to Mendel's original principles:

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

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Objective 19a

a) 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:

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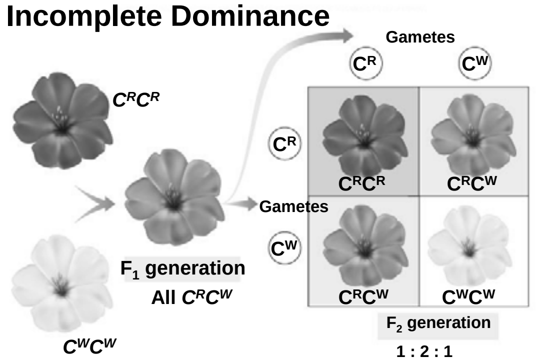
Incomplete Dominance



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Objective 19b

b) 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 19c

c) 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 19c

- 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 19c

- 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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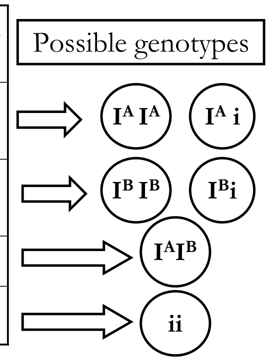
Objective 19c

Antigens present	Blood Type
galactosamine only	A
galactose only	B
both	AB
neither	O



I^A and I^B are codominant

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Objective 19c

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

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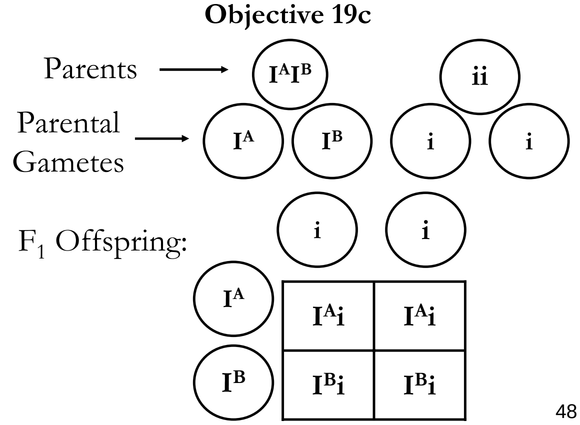
Objective 19c



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Objective 19d

d) 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, such as height in humans, often show continuous variation, rather then a few discrete forms:

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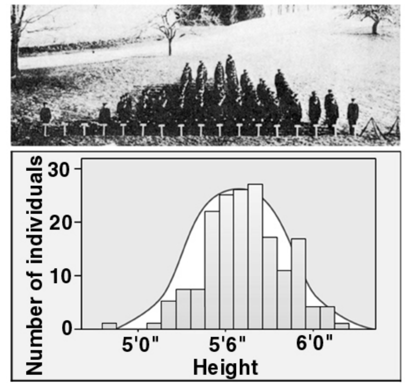
Continuous Variation



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Objective 19e

e) 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.

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Objective 19e

- 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:

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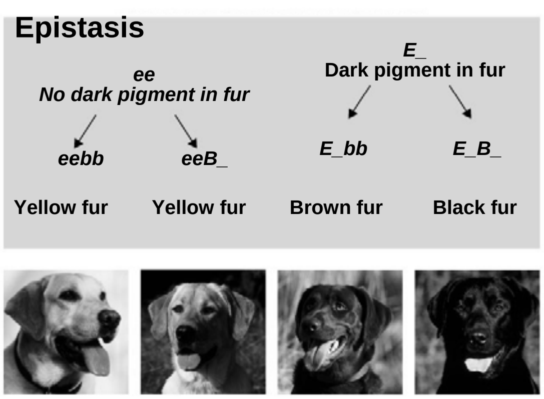

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Objective 19f

f) 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.

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Objective 19g

g) 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.

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Environmental Effects on an Allele

Arctic Fox in Winter

Arctic Fox in Summer

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Objective 19g

- 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.

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