Patterns of Inheritance

Introduction

- Close observation of breeding organisms and their offspring show patterns in the inheritance of characteristics.
- The trait variant most common in nature is referred to as the ***wild type***.
- Pattern of inheritance can be explained by the behavior of chromosomes during meiosis and fertilization.

Mendel's Principles

- The ancient Greeks believed in pangenesis, the idea that particles governing the inheritance of each characteristic collect in eggs and sperm and are passed on to the next generation.
- But many including Aristotle, realized there were problems with this idea: The potential to produce characteristics is inherited, not pieces of characteristics themselves. Reproductive cells are not changed by the development or activity of other cells.

- Based on artificial breeding, nineteenth-century observers believed in the "blending" hypothesis, in which the characteristics from both parents blend in the offspring.

Painting of Mendel

Experimental Genetic began in an abbeys garden

•Mendel studied peas because they offered an advantage over other organisms. Peas grow easily, they have relatively **short life spans**, have **numerous and distinct characteristics**, and the **mating of individuals can be controlled so that the percentage of offspring can be known for certain**.

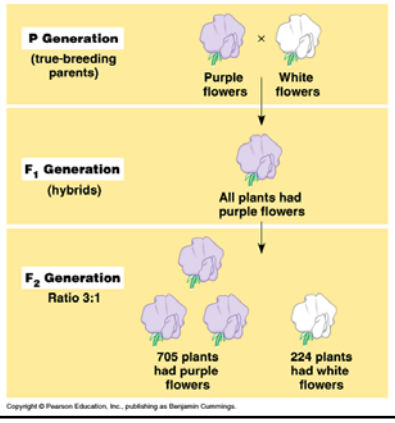
•Mendel's paper, published in 1866, argued that there are discrete, heritable factors (what we call genes) that retain their individuality when transmitted from generation to generation.

•Mendel could intentionally self-fertilize a flower by covering it with a bag, or cross fertilize two different plants by dusting the carpels of one with the pollen of another.

• By continuous self-fertilizations for many generations, Mendel developed breeds of plants that bred true (continued to show characteristic when self-fertilized) for each of the characteristics he followed. He found seven characteristics, each of which came in two distinct forms.

•Mendel developed two principles based on two types of experiments. In one type (monohybrid crosses), he hybridized true-breeding plants for each of the two forms of a characteristic. In a second type (dihybrid and trihybrid crosses), he hybridized plants that combines two or more of the seven characteristics.

•In these experiments, the true breeding parents are the **P** (parental) generation, their hybrid offspring is the **F_1** (first filial) generation, and the offspring of mating two **F_1s** is the **F_2** (second filial generation).



Mendel's principle of segregation describes inheritance of a single characteristic

•**Principle of segregation:** pairs of genes segregate during gamete formation; the fusion of gametes at fertilization pairs genes once again.

•Mendel conducted a monohybrid cross with flower color . The results of this experiment were: out of 929 **F_2** offspring, 705 were purple, and 224 were white.

Note: The proportions are not exactly ¾ and ¼ because mating involves probabilities.

Mendel developed four hypotheses:

•There are alternative forms of genes, the units that determine heritable characteristics. These alternative forms are called **alleles**.

•For each inherited characteristic, an organism has two genes, one from each parent. They maybe the same allele or different alleles.

•A sperm or egg carries only one allele for each characteristic because the allele pairs segregate from each other during gamete production.

•When the two alleles are different, the one that is fully expressed is said to be **dominant** and the one that is not noticeably expressed is said to be **recessive**.

•**Convention for alleles**: *P*, the dominant (purple) allele, and *p*, the recessive (white) allele. **P** generation: *PP* x *pp*; their gamete: *P* and *p*; $\mathbf{F_1}$ generation: *Pp*.

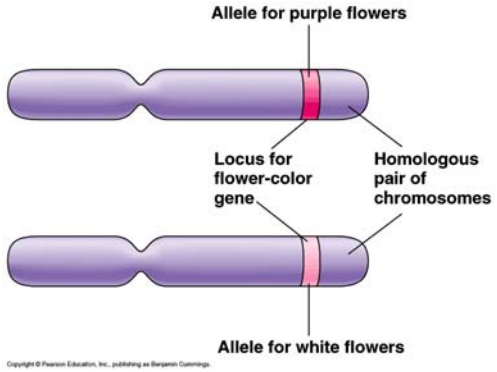
•**Homozygous dominant**, **homozygous recessive**, and **heterozygous** refer to the **genotypes**. The **phenotypes** are what we see.

•The Punnet square is used to keep track of the gametes (two sides of the square) and offspring (cells within the square).

Homologous chromosomes bear two alleles for each characteristic

•Although Mendel knew nothing about chromosomes, our knowledge of chromosome arrangements (in homolgous pairs) strongly supports the principle of segregation.

•Alleles of a gene reside at the same **locus** on homologous chromosomes.



The principle of independent assortment is revealed by tracking two characteristics

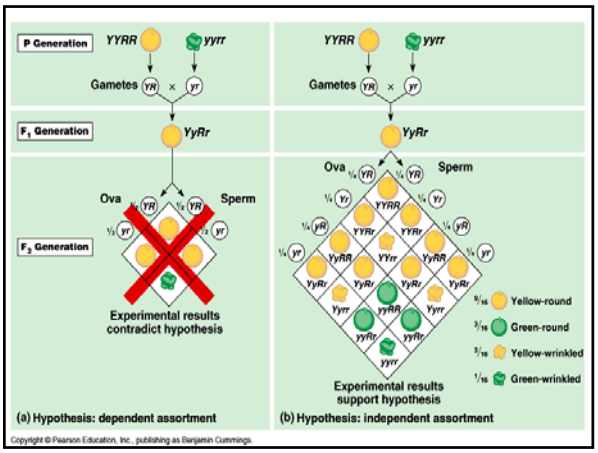
Principle of independent assortment: each pair of alleles segregates independently during gamete formation.

•**Example procedure:** Breed two strains true, each exhibiting one of the two forms of two characteristics. For example round yellow seeded plants (*RRYY*) and wrinkled green seeded plants (*rryy*). Hybridize these as the **P** generation, resulting in hybrid offspring (**F_1**:*RrYy* x *RrYy).*

Note: Each of these individuals produces the same four gametes: *RY*, *Ry*, *rY*, *ry*. Taking one gamete from each individual means there are $4^2 = 16$ possible genetic combinations.

•Results: The F_1 generation exhibits only dominant phenotypes (this is expected). The F_2 generation exhibits a phenotypic ratio of 9:3:3:1 (round yellow: round green: wrinkled yellow: wrinkled green).

Note: 9 + 3 + 3 + 1 = 16, the same as the number of possible gametic combinations. That the phenotypic ratio adds up to the number of gametic combinations serves as a check for the result of a cross.



Geneticists use the testcross to determine the unknown genotypes

•A testcross involves crossing an unknown genotype expressing the dominant phenotype with the recessive phenotype (by necessity, homozygous).

•Each of the two possible genotypes (homozygous or heterozygous) gives a different phenotypic ratio in the F_1 generation. Homozygous dominant gives all dominant. Heterozygous gives half recessive, half dominant.

NOTE: This technique uses phenotypic results to determine genotypes.

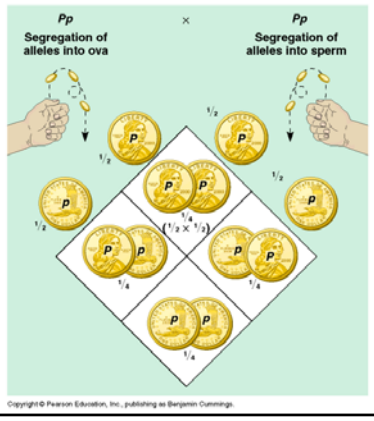
Mendel's principles reflect the rules of probability

•Events that follow probability rules are independent events; that is one such event does not influence the outcome of later such events. If you flip a coin four times and get four heads, the probability for tails on the next flip is still 1/2.

•The probability of two events occurring together is the product of the probabilities of the two events occurring apart (the rule of multiplication).

• Thus, when studying how alleles of two (or more) genes that segregate independently behave, use the probabilities of how they behave individually.

NOTE: The probability of a recessive phenotype occurring in a monohybrid cross is 1 out of 4. The probability of two recessives occurring together in a dihybrid cross ¼ x ¼ , or 1 out of 16 (recall 9+3+3+1=16). In a trihybrid cross, as mentioned, the probability of triple recessive is 1 out of 64.



•If there is more than one way an outcome can occur, these probabilities must be added, as in the case of determining the chances of heterozygous mixtures (the rule of addition).

Genetic traits in humans can be tracked through family pedigrees

•A pedigree shows a family's pattern of inheritance for a specific trait.

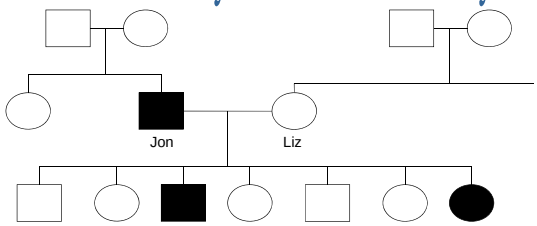
•In a pedigree, each person has a number and each generation is represented by a Roman numeral. In this way, each individual is identified by a Roman numeral and an Arabic numeral.

•Females are represented by circles and males by squares.

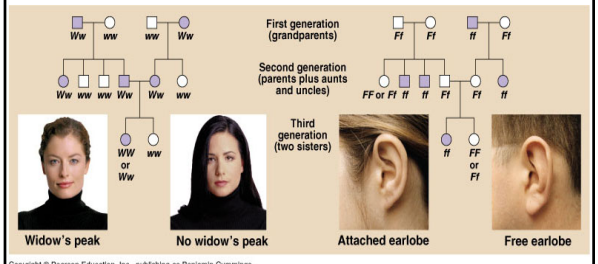
•Shaded symbols indicate individuals who have the trait being studied.

•By applying Mendel's principles one can deduce the information on the chart from the patterns of phenotypes.

Deafness in a Family from Martha's Vineyard



Congenital Deafness: Assuming Jon inherited his deafness from his parents , the only explanation is that his deafness is caused by a recessive allele because neither of his parents were deaf. Because some of his children were deaf, his wife, Liz must be a carrier. From this we know all the hearing children are carriers.



Many inherited disorders are controlled by a single gene

•Over 100 known genetic traits are attributable to a single gene locus and show simple Mendelian patterns of inheritance.

•Many human characteristics are though to be determined by simple dominant-recessive inheritance, and sometimes the ratio of dominant-to-recessive phenotypes exhibits a Mendelian ratio.

•Most disorders are caused by recessive alleles and vary in the severity of the expressed trait.

•The vast majority of people afflicted with recessive disorders are born to normal, heterozygous parents.

• Cystic fibrosis is the most common lethal genetic disease in the U.S..

•Most genetic diseases of this sort are not evenly distributed across racial and cultural groups because of the prior and existing reproductive isolation of various populations.

•Laws forbidding inbreeding may have arisen from observations that reproductive attempts in such marriages more often resulted in miscarriages, stillbirths, and birth defects.

•Some disorders are caused by dominant alleles. These disorders vary in how deadly they are. Some are nonlethal handicaps, some are lethal in the homozygous condition, and some are intermediate in severity.

•***Achondroplasia***, a type of dwarfism, is lethal in the homozygous condition; individuals who express the trait are heterozygous.

•Other conditions attributable to dominant alleles are lethal only in older adults, so the allele can be passed to children before it is realized that the parent has the condition.

i.e.- Huntigton's Disease

Recessive Disorders

•Albinism
•Cystic Fibrosis
•Galactosemia
•PKU
•Sickle Cell
•Tay-sachs

Dominant Disorders

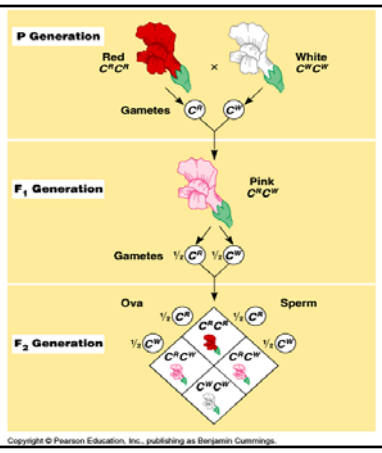
•Achondroplasia
•Alzheimer's (1 type)
•Huntington's Disease
•Hypercholesterolemia

•The principles and techniques of genetics can be used to make informed decisions about bearing children. (**genetic counseling**)

Incomplete dominance results in intermediate phenotypes

•Incomplete dominance describes the situation where one allele is not completely dominant in the heterozygote; the heterozygote usually exhibits characteristics exhibits characteristics intermediate between both homozygous conditions.

i.e.- snapdragon color



Hypercholesterolemia

• Normal individuals, **HH**, have normal amounts of LDL receptors

• **hh** individuals have no receptors and 5X the amount of blood cholesterol.

• **Hh** individuals have half the number of receptors and 2X the amount of blood cholesterol.

Many genes have more than two alleles in the population

•The ABO blood groups in humans follow this pattern, in which individuals can have two alleles from a set of three possible alleles.

•These blood-type alleles code for two carbohydrates (or absence of any carbohydrates) on the surface of red blood cells (a total of three alleles). There are six possible genotypes and four possible phenotypes.

•When blood is transfused, recipients develop antibodies for the types of carbohydrate on the donor red blood cells that the recipient lacks.

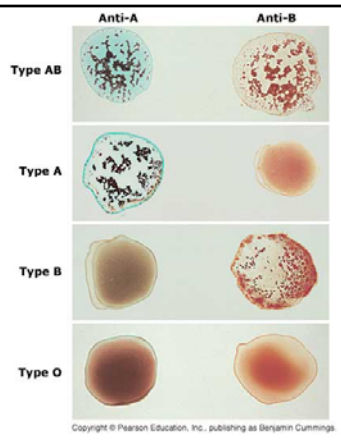
•Type **O** (universal donor) has neither carbohydrate and can receive no other type. Type **AB** (universal recipient) has both carbohydrates and can receive any type. Type **A** has carbohydrate A and can receive A or O. Type **B** has carbohydrate B and can receive B or O.

•Blood types can be used can be used to disprove or suggest parentage in paternity suits.

(a) Phenotype (blood group)	(b) Genotypes (see p.258)	(c) Antibodies present in blood serum	(d) Results from adding red blood cells from groups below to serum from groups at left			
			A	B	AB	O
A	$I^A I^A$ or $I^A i$	Anti-B				
B	$I^B I^B$ or $I^B i$	Anti-A				
AB	$I^A I^B$	—				
O	ii	Anti-A Anti-B				

	I^A	I^B
i	$I^A i$	$I^B i$
i	$I^A i$	$I^B i$

	I^A	i
I^B	$I^A I^B$	$I^B i$
i	$I^A i$	ii



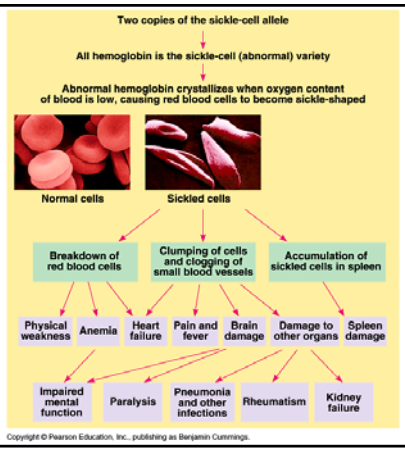
A single gene may affect many phenotypic characteristics

•This common situation is known as **pleiotropy**.

•An example is the inheritance of an allele that codes for abnormal hemoglobin and, in the homozygous condition, causes **sickle-cell disease**.

•The normal and abnormal alleles are **codominant**, so heterozygous individuals (carriers) can exhibit some symptoms.

•The incidence of the allele is relatively high in individuals of African descent (1 in 10 African Americans are heterozygous), because sickle cell carriers are somewhat protected from malaria.

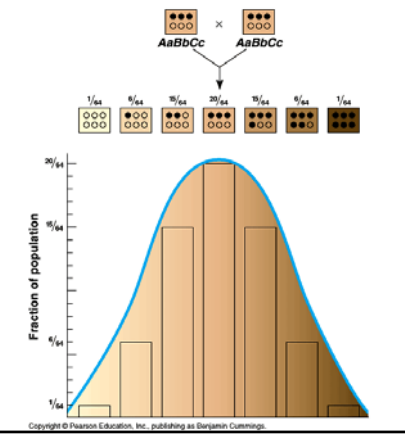


A single characteristic may be influenced by many genes

•This situation is known as **polygenic inheritance**.

•Skin pigmentation is just such a phenotypic characteristic whose underlying genetics have not been completely determined.

•Figure 9.16 in the text is a hypothetical, showing the phenotypic outcome of mixtures of three genes, each with two alleles coding for "additive units," which produce the overall characteristic.
i.e.- **aabbcc**- very light
AABBCC- very dark
AaBbCc- intermediate shade



Genes on the same chromosome tend to be inherited together

•Linked genes are located close together on the same chromosome.uh

•The inheritance described does not follow the pattern described by the principle of independent assortment because the two genes are normally inherited together.

i.e.- purple flowers and long pollen grains in pea plants. (3:1 rather than 9:3:3:1)

Sex-linked disorders affect mostly males

•Examples of such characteristics are **red-green color blindness**, a type of **muscular dystrophy**, and **hemophilia**.

•Because the male has only one X chromosome, his recessive X-linked characteristic will always be exhibited.

•Most known sex-linked traits are caused by genes (alleles) on the X chromosome.

•When these traits are recessive (most are), males express them because they have only one X. Females who have the allele are normally carriers and will exhibit the condition only if they are homozygous.

•Males cannot pass sex-linked traits to sons (who get a Y from their father).

	X^R	**Y**
X^R	X^RX^R	X^RY
X^r	X^RX^r	X^rY