

GENETICS

Basic Concepts

A. Organismal Reproduction

- One of the most important requisites of all life, from the earliest life forms to present-day organisms, is **reproduction**
- Characteristics or **traits** of organisms must be passed on during reproduction

B. Cellular Reproduction

- Life as we know it is based on the cell, the basic unit of life
- Cell theory states all organisms are made up of cells and come from cells

C. DNA

- DNA (**d**eoxyri**n**ucleic **a**cid) is the **molecule of inheritance** in ALL cellular forms of life

D. Chromosomes

- Eukaryotic** cells possess nuclear DNA with structural and enzymatic proteins, forming **chromatin**, which is visible as **chromosomes** during parts of the cell cycle
- Prokaryotic** cells possess simpler DNA
- Sexually reproducing organisms typically have pairs of **homologous chromosomes** (look-alike chromosomes)

E. RNA

- RNA (**r**ibonucleic **a**cid) is found in several forms, most of which are used in protein synthesis
- RNA is the **molecule of inheritance** in some viruses, which are not cell-based life forms

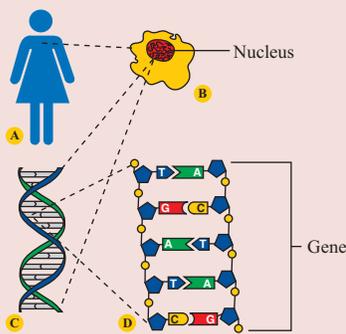
F. Genes

- Functional unit of inheritance and **basis for most traits**
- Located at **loci**, or specific positions, on DNA; to be preserved and transmitted
- Control biological processes through production of proteins and RNA

- Genotype** refers to the *genetic composition* of the organism

- Phenotype** refers to the *observable inherited traits* (e.g., physical, behavioral, physiological characteristics); based on the inherited genotype

Genes Form Basis of Inheritance



G. Ploidy

- Homologous chromosome pairs have the same loci, thus genes
- When both chromosomes are present, for each gene there are two representatives; this is represented by the symbol **2n** or **diploid** condition
- When only half of each homologous chromosome pair is present, such as in **gametes**, this is represented by the symbol **n** or **haploid**

H. Alleles

- Alternate forms of the same gene that could occupy the same locus (e.g., brown versus blue eye color)

- Homologous chromosomes** possess two representatives of each gene (i.e., 2n)
- Homozygous** refers to the diploid condition where **both alleles of the genotype are identical** (e.g., AA, aa)
- Heterozygous** refers to the diploid condition where **both alleles of the genotype are different** (i.e., Aa)
- Dominant alleles** form a phenotypic expression regardless of the other allele on the matched chromosome of the homologue (e.g., "AA" or "Aa" genotypes will both express the phenotype designated by the "A" allele)
- Recessive alleles** fail to form a phenotypic expression unless the other allele on the matched chromosome is also recessive (e.g., "aa" genotype is the only way for the phenotype designated by the "a" allele to be expressed, assuming no other gene pairs influence inheritance [see **epistasis** discussion in **Gene Action Categories**, page 2])
- Additional types of allelic interactions will be discussed in subsequent sections
- Determining gamete types:** Assuming there are no mutations, alleles present in gametes are determined by the diploid genotypes of parents
 - For **homozygous genotypes**, *haploid gametes will be identical for the given traits* (i.e., AA individual would produce "A" gametes only; AAbb individual would produce "Ab" gametes only)
 - For **heterozygous genotypes**, *haploid gametes will be different for the given traits* (i.e., Aa individual would produce "A" & "a" gametes; AaBb individual would produce "AB, Ab, aB, ab" gametes—assuming two traits are unlinked [see **Independent Assortment & Dihybrid Crosses**, page 2])

Mendelian Genetics

A. Gregor Mendel (1822–1884)

- An Austrian monk who, through his love and interest in nature, developed the basic ideas of genetics long before chromosomes and genes (i.e., molecular biology) were discovered
 - He developed his ideas by studying plants; in particular, his most famous work involved crosses with **pea plant varieties**
- His results and interpretations contrasted with a prevailing (at that time) theme of inheritance called "**blending**"—the concept that *inherited traits mixed to create a composite characteristic in offspring*

B. Mendel's Genetics Laws

1. Segregation of Alternate Factors & Monohybrid Crosses

- Specifically, Mendel discovered that with certain traits, there were individual plants which, if only crossed with other plants just like them, would almost always produce the exact same phenotype
 - These individuals were called **true-breeders**
 - We now call this condition **homozygous**
- He also found that some individuals with similar appearance, when crossed, would not have all offspring of the same kind
 - We now call this condition **heterozygous**
- Mendel decided to systematically do **single-trait crosses** to determine the causes for the previously stated observations
- Specifically, a **parental generation (P)** initiated these experimental crosses by using two true-breeding pea plants for opposite phenotypes (e.g., purple versus white flowers)
- Offspring** from this cross (**F₁**) all showed only one of the traits (e.g., purple flowers), and this trait was called the **dominant trait**
- Traits from the P generation "**did not blend**" in these F₁ individuals
- F₁ individuals, the **hybrids**, were cross-pollinated—the **monohybrid cross**—to produce F₂ individuals
- 3/4 of the F₂ individuals expressed the dominant trait, while 1/4 expressed the trait of the other P parent (e.g., white) that had not been expressed in the F₁ generation—this latter trait was the **recessive form**
- The expected **phenotypic ratio** of the F₂ individuals in **monohybrid** crosses would be **3:1**
- The expected **genotypic ratio** of the F₂ individuals in **monohybrid** crosses would be **2:1:1**
- The diagram at right, called a **Punnett square**, summarizes results of a single-trait cross similar to those done by Mendel on pea plants and other organisms:

Mendel's 1st Law: Segregation of Alternate Factors

P generation	Gray female (GG)	Normal male (gg)	
Gametes produced by P generation	G	g	
F ₁ generation	All Gg		
	Dominant G masks recessive g		
	Gametes produced by F ₁ generation	G	g
All Gg	G	GG	Gg
	g	Gg	gg
		F ₂ Generation	

- Mendel concluded there had to be some physical entities or "factors" passed on by each parent of a cross
 - We now know these to be **genes**
 - He also concluded that these factors came in **pairs**, which then became **unpaired** (in the production of *gametes*, which occurs during *meiosis*) and **recombined** during **fertilization**
 - The two P generation individuals had the factors in *alternate forms* called **alleles** (e.g., purple versus white flowers)

- iv. Each of these true-breeding parent plants had a pair of identical factors, but their gametes had only one
- v. Thus, F₁ individuals were hybrids genotypically, but only expressed the dominant phenotype

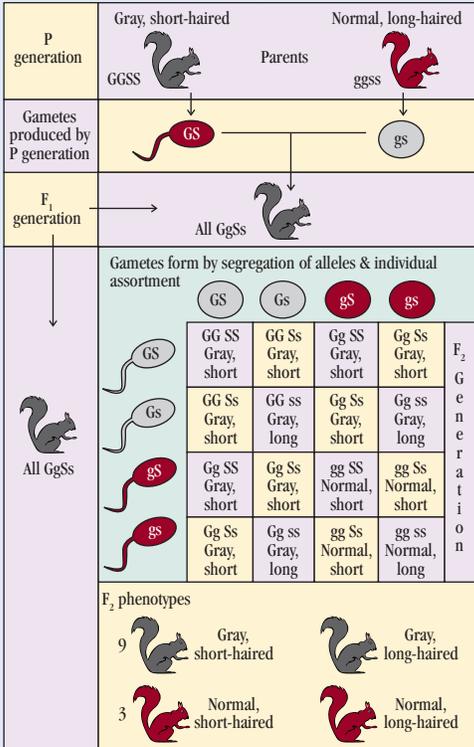
m. **Monohybrid Cross:** Once Mendel realized the F₁ individuals were genotypic “hybrids,” he predicted the recessive trait that “disappeared” would reappear if:

- i. F₁ hybrid individuals were crossed to produce F₂ offspring
- ii. The results summarized in “h” above confirmed his predictions

2. Independent Assortment & Dihybrid Crosses

- a. Mendel continued his crossing experiments by looking at **multiple traits** simultaneously
- b. P generation, consisting of two true-breeding parents of different forms (phenotypes) for **two traits**, were crossed, producing F₁ individuals
- c. The F₁ genotypic hybrids for both traits were crossed—the **dihybrid cross**—producing F₂ individuals
- d. 9/16 of the F₂ individuals expressed both dominant traits; 3/16 expressed 1 dominant trait, and 1 recessive trait; 3/16 expressed the opposite dominant trait, and the opposite recessive trait; 1/16 expressed both recessive traits
- e. The expected **phenotypic ratio** of the F₂ individuals in dihybrid crosses would be **9:3:3:1**
- f. The expected **genotypic ratio** of the F₂ individuals in dihybrid crosses would be **1:1:1:1:2:2:2:2:4**—a total of 9 genotypes
- g. The following Punnett square summarizes results of two-trait crosses, similar to those done by Mendel:

Mendel’s 2nd Law: Independent Assortment



- h. Mendel concluded statistically that these results occurred because the *alleles for one trait did not affect the inheritance of alleles for the other trait*, which is **independent assortment**
- i. Special note: Mendel did not observe independent assortment for all traits studied [see **Mendel’s Ratios & Beyond**, “C” on this page, for more about **gene linkage**]

3. Trihybrid Crosses & Beyond

- a. Tracking three or more traits simultaneously is possible; the following summarizes such crosses:
P = AABBCC x aabbcc (true breeders crossed)
F₁ = AaBbCc x AaBbCc (trihybrid individuals crossed)
F₂ = 27:9:9:9:3:3:3:1 (phenotypic ratio)
- b. The following Punnett square summarizes results of Mendel’s three-trait crosses—specifically, the F₂ individuals produced from the F₁ trihybrid individuals:

Trihybrid Cross								
	ABC							
ABC	AABBCC							
ABc	AABBCc							
AbC	AABbCC							
Abc	AABbCc							
aBC	AaBBCC							
aBc	AaBBCc							
abC	AaBbCC							
abc	AaBbCc							

c. **Probability rules** can be used to calculate genotypes and phenotypes, in place of using Punnett squares (especially useful in multiple-trait crosses)

i. **Addition Rule:** The occurrence of mutually exclusive events equals the sum of their individual probabilities; that is, calculate probabilities associated with the dominant and recessive alleles as demonstrated in a monohybrid cross:

- AA = 1/4, Aa = 1/2, aa = 1/4
- For example, in a monohybrid cross, the chance of a **dominant phenotype** is equal to 1/4 (AA) + 1/2 (Aa) = 3/4

ii. **Multiplication Rule:** The probability of independent events occurring simultaneously is equal to the product of their individual probabilities

- For example, the probability of being AABbcc = (1/4) x (1/2) x (1/4) = 1/32 OR 2/64 [see Punnett square above]

d. **Branch (Fork) Diagrams** are alternatives to Punnett squares; multiplication rule used to calculate genotypic and phenotypic ratios

e. Additional mathematical relationships associated with multiple-trait crosses

- i. n = number of heterozygous gene pairs
- ii. 2ⁿ = number of different gametes formed
- iii. 3ⁿ = number of different genotypes formed
- iv. 2ⁿ = number of different phenotypes formed

4. **Back Cross:** A cross of an F₁ individual (Aa) with either of the two P generation individuals (AA or aa)

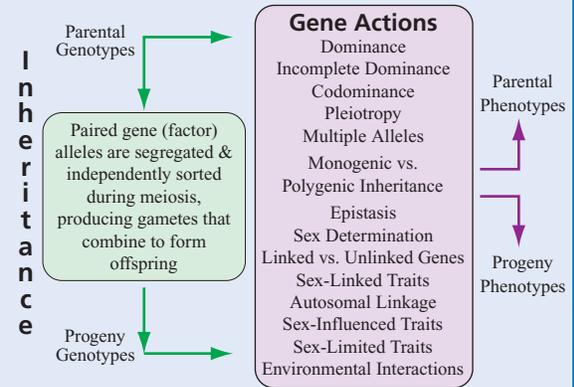
5. **Test Cross:** A cross of an individual having a dominant phenotype (but unknown genotype—e.g., AA or Aa) with an individual that is homozygous recessive (aa)

- a. If the recessive phenotype shows up in approximately half of the offspring, the unknown genotype is determined to be a **heterozygote**

C. Mendel’s Ratios & Beyond

- 1. Mendel’s work paved the way for the most basic understanding of inheritance; however, future discoveries revealed that many traits are inherited in ways much more complex than those demonstrated in the basic monohybrid and dihybrid crosses
- 2. Thus, 3:1 and 9:3:3:1 phenotypic ratios are uncommon in nature

3. The following diagram summarizes many types of gene actions, each of which will be discussed in greater detail in the sections that follow



D. Gene Action Categories

- a. **Dominance:** One allele dominates or masks the effects of the other allele(s)
- b. **Incomplete Dominance:** Neither allele is expressed fully; in such cases, *phenotypes are “blended”*
- c. **Codominance:** Both alleles are expressed fully (**NOTE:** It is frequently difficult to distinguish this pattern from incomplete dominance)
- d. **Pleiotropy:** One gene affects several phenotypes
- e. **Multiple Alleles:** Three or more alleles for a gene are present within a population (although diploid individuals can only have two at a time)
- f. **Monogenic versus Polygenic Inheritance:** Traits based on a single gene versus traits based on multiple genes
- g. **Epistasis:** One gene alters the effect of another gene
- h. **Sex Determination:** For many organisms, special chromosomes have genes that determine gender; in some, such as sea turtles and alligators, environmental factors, such as the temperature at which eggs develop, determine gender
- i. **Linked versus Unlinked Genes:** Genes on the **same chromosome** are **linked**; genes on **different chromosomes** are **unlinked** and **assort independently**
 - i. **Gene Mapping:** Recombinant progeny (involving crosses) can be used in some organisms to map gene loci; molecular techniques are used for many species, including humans (e.g., **Human Genome Project** [also see **structural genomics** discussion in **Molecular Genetics**, page 5])
- j. **Sex Linkage:** In humans, genes found on the X or Y chromosomes (e.g., color blindness)
- k. **Autosomal Linkage:** Multiple genes found on non-sex chromosomes
 - 1. **Sex-Influenced Traits:** Same genotype expressed differently in males versus females (e.g., baldness in humans)

- m. **Sex-Limited Traits:** Same genotype expressed only in one sex; suppressed in the opposite sex (e.g., beard development and breast development in humans)
- n. **Chromosomal Non-Disjunctions:** During meiosis, chromatids and/or homologous chromosomes may fail to separate, triggering alterations in phenotypic expressions of genotypes
 - i. **Aneuploidy:** Abnormal number (too few/too many; missing pieces/extra pieces) of chromosomes
 - o. **Polyploidy:** Presence of more than two sets of chromosomes (e.g., $3n$ = triploid)
- p. **Environmental Effects:** Phenotypes that are affected by non-genetic, environmental factors (e.g., differential pigment development in Siamese cats) based on temperature; cooler body areas have heavier melanin deposition [see **sex determination**, item “h” in this list]

E. Human Genetics

1. We know more about the genetics of many organisms than that of humans, mostly because there are fewer ethical issues and shorter generation times for non-human organisms
2. The Human Genome Project has helped in the discovery of genes and their functions through molecular studies [see **Molecular Genetics** section, page 5]
3. The inheritance patterns of some human traits have been worked out (using mostly Mendelian Genetics) and are summarized in the table that follows:

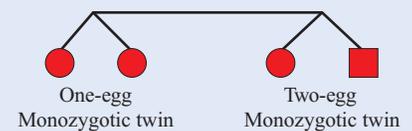
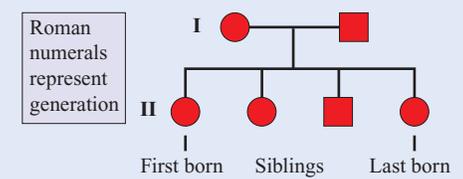
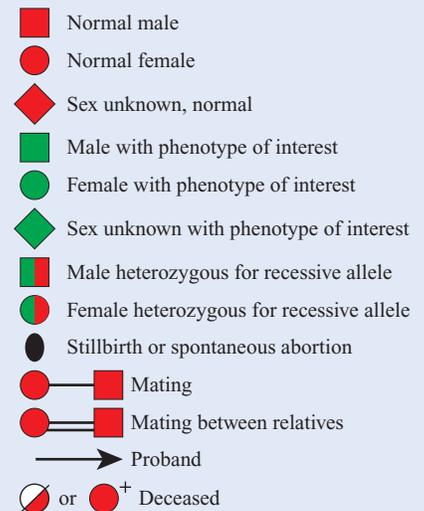
Human Traits & Known Inheritance Patterns

Name of Trait	Phenotypes	Mode of Inheritance
ABO Blood Groups	Type A, B, AB, O	<u>Autosomal Codominant</u> – Multiple Alleles: Type A = AA or I _A I _A ; AO or I _A i Type B = BB or I _B I _B ; BO or I _B i Type AB = AB or I _A I _B Type O = OO or ii
Achondroplasia	Dwarfism	<u>Autosomal Dominant:</u> Aa = dwarf, aa = normal (AA is lethal)
Albinism	Lack of pigmentation in eyes, hair, skin	<u>Autosomal Recessive:</u> A_ = normal, aa = albino
Color Blindness	Cannot distinguish red or green	<u>Sex-Linked Recessive:</u> X ^C X ^C or X ^C X ^c = normal-vision female X ^C Y = normal-vision male X ^c X ^c = color-blind female X ^c Y = color-blind male
Cystic Fibrosis	Hypersecretion of mucus in lungs	<u>Autosomal Recessive:</u> C_ = normal, cc = disease
Dimples	Dimple(s) in cheek(s)	<u>Autosomal Dominant:</u> D_ = dimples, dd = no dimples
Ear Lobes	Free vs. attached	<u>Autosomal Dominant:</u> D_ = free lobes, dd = attached lobes
Eye Color	Blue → dark brown	<u>Autosomal Incomplete Dominant:</u> BB = dk. brown, Bb = lt. brown, bb = blue, model with 3 Genes? Recent studies suggest “NO” specific eye color genes exist
Freckles	Freckles vs. no freckles	<u>Autosomal Dominant:</u> F_ = freckles, ff = no freckles
Hairy Ears	Hair on ear edge (pinna)	<u>Y-Linked Dominant:</u> XY ^H = hairy-eared male, XY ^h = normal male
Hairline Shape	Widow’s peak vs. straight	<u>Autosomal Dominant:</u> W_ = widow’s peak, ww = straight line
Height	Variable height	<u>Polygenic:</u> aabbccddeeff = shortest, AABBBCCDDEEFF = tallest
Hemophilia	Blood clotting impairment	<u>Sex-Linked Recessive:</u> X ^H X ^H or X ^H X ^h = normal female X ^h X ^h = hemophiliac female X ^H Y = normal male X ^h Y = hemophiliac male
Huntington’s Disease	Mental decay	<u>Autosomal Dominant:</u> H_ = disease, hh = normal
Muscular Dystrophy “Duchenne”	Muscle weakening & loss of coordination	<u>Sex-Linked Recessive:</u> X ^D X ^D or X ^D X ^d = normal female X ^d X ^d = dystrophic female X ^D Y = normal male X ^d Y = dystrophic male
Pattern Baldness	Receding hairline	<u>Sex-Influenced:</u> BB = bald male, thin-hair female Bb = bald male, full-hair female bb = full hair in both sexes
Phenylketonuria (PKU)	Missing enzyme for phenylalanine metabolism	<u>Autosomal Recessive:</u> P_ = normal (results in mental impairment), pp = PKU
Polydactyly	Extra toes & fingers	<u>Autosomal Dominant, Incomplete Penetrance:</u> P_ = extra toes or fingers (but may be normal) pp = normal # of toes & fingers
Rh Blood Groups	Rh factor on red blood cells (RBC)	<u>Autosomal Dominant:</u> R_ = Rh-positive, r = Rh-negative
Sickle-Cell Anemia	Defective RBC blood disorder	<u>Autosomal Incomplete Dominant:</u> SS = anemia, Ss = carrier, ss = normal
Skin Pigmentation	Dark to light	<u>Polygenic:</u> aabbcc = lightest → AABBBCC = darkest
Tay-Sachs Disease	Nervous degeneration	<u>Autosomal Recessive:</u> T_ = normal, Tt = carrier, tt = disease
Tongue Folding	Fold vs. not fold	<u>Autosomal Dominant:</u> F_ = fold, ff = unable to fold tongue
Tongue Rolling	Roll vs. not roll	<u>Autosomal Dominant:</u> R_ = roll, rr = unable to roll tongue

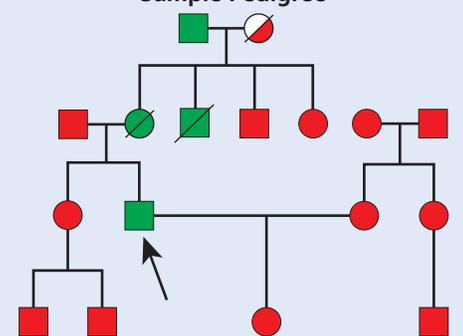
4. **Human Pedigree:** Studying inheritance patterns of humans is complex both biologically and ethically; thus, much of what we know is based on looking at family histories or trees (**pedigree analysis**)

- a. Specifically, phenotypes of all known family members from as many generations as possible are assembled; this is especially important when attempting to trace the sources/causes of **genetic disorders**
 - i. **Proband** refers to the first person for whom a particular genetic condition has been diagnosed: If this is a male, he is called the **propositus**; if this is a female, she is called the **proposita**
 - ii. The diagnosis and identification of the proband individual serves as the basis for determining the genetic basis of the condition through the use of standardized diagrams
 - iii. Following is a chart illustrating some standard symbols used in pedigrees and a sample pedigree:

Human Pedigree Symbols



Sample Pedigree



Sample Genetics Problems

Please refer to the **Human Traits & Known Inheritance Patterns** table [see page 3] for assistance with problems involving human traits; attempt to work out the problems on a blank sheet of paper before viewing the solutions to each problem, which are shown in the next section [see **Solutions to Sample Genetics Problems**, pages 4–5]; in crosses tracking more than one trait simultaneously, assume multiple traits are unlinked unless stated otherwise

- Orange and black fur in domestic cats are sex-linked, with the gene locus on the X chromosome; cats have similar sex determination patterns as humans; the two fur-color alleles display codominance, with heterozygous cats displaying a calico pattern (i.e., separate patches of black and orange fur; a calico female is mated to a black male):
 - What is the probability that a female kitten will be a calico?
 - What is the probability a male kitten will be a calico?
- There is a gene that affects peapod color and has two alleles: $G = \text{yellow}$ and $g = \text{green}$; there is another gene that affects peapod shape and has two alleles: $W = \text{round}$ and $w = \text{wrinkled}$:
 - Mendel's original crosses (P generation) would have consisted of which two genotypes?
 - List the possible types of gametes produced by these original parent plants
 - If gametes from question "b" are joined in fertilization, determine the genotype(s) of the offspring
 - If the individuals of question "c" are crossed with each other, how many genotypes will be possible in the offspring?
 - Determine the phenotypic ratios of the offspring produced in question "d"
- A certain hypothetical species called an EWOK has a gene that controls "fur" color—a dominant allele causes blue fur, and a recessive allele causes red fur; another gene controls "ear" length—a dominant allele causes long ears, and a recessive allele causes short ears; these two genes are linked, and a cross is performed between an individual homozygous for both dominant traits and an individual homozygous recessive for both traits:
 - Determine the phenotypes(s) of all possible offspring
 - How many types of gametes can be produced by the offspring listed in question "a"?
 - What would be the possible phenotypes of the offspring in a test cross involving the offspring of question "a"? (Do a test cross on the offspring listed in question "a"); ASSUME NO CROSSING OVER
 - Do the same thing here as you did in question "c," but now give the possible phenotypes ASSUMING CROSSING HAS OCCURRED
- Jenna has type A blood and does not exhibit symptoms of diabetes; her husband, Stephan, has type B blood and is diabetic; Stephan and Jenna have a male child, Oscar, who has type O blood and, like his father, has diabetes; based on the information given, answer the following questions:
 - Name the genotypes of both parents and Oscar
 - What is the probability that their next child will have the same genotype and phenotype as Oscar's?

- A genetics marriage counselor has a couple, Carly and Cedric, both with no hemophilia symptoms, seeking professional advice; Carly says that both her parents are normal, but she has a brother with hemophilia; Cedric, the man she will marry, is normal but knows one of his cousins has hemophilia:
 - What is the probability of disease in children they might have?
- In cattle, when red and white cattle are mated, the offspring are roan (a mixture of red and white fur); for another trait in cattle, hornless is dominant to possessing horns; white hornless cattle and red horned cattle are crossed:
 - F_1 individuals would have what genotype(s)?
 - If F_1 individuals are crossed, how many would be expected to be roan and hornless?
 - If F_1 individuals are crossed, how many would be expected to be white and horned?
- Three siblings, a brother and two sisters, are examined for color-blindness; the examiner discovers that the boy and one sister are color-blind, while the other sister has normal color vision:
 - Determine the genotype and phenotype of each parent
- Diabetes, a recessive trait in humans, occurs in 10% of a certain population; a normal female and a normal male in this population have a child who is diabetic:
 - What is the probability that their next child will be a diabetic?
- A "hypothetical" recessive allele in humans causes feet to grow backward, while the normal dominant allele causes feet to grow forward; another gene controls foot length, and its alleles display incomplete dominance, so when a person is heterozygous for foot-length alleles, he/she will have normal-sized feet, but when homozygous for either of the two foot-length alleles, the person will be either big-footed or small-footed; a cross occurs between a backward-pointing, large-footed male and a homozygous forward-pointing, normal-size-footed female:
 - The female can produce how many types of gametes as far as these traits are concerned?
 - Determine all possible phenotypes of the offspring
 - In the original cross, if the male parent had backward-pointing feet of normal size, how many different phenotypes in the offspring would be possible?
 - In the original cross, if the female parent had normal-size feet but was backward-pointing, how many different phenotypes in the offspring would be possible?
 - In the cross with the female in question, "d," will any offspring have forward-pointing feet?
- Sheri has type A blood and normal color vision capabilities; her mother was type O and also had normal color vision, while her father was type AB and color-blind; Sheri recently married Randy, who had identical phenotypes for both traits as those of her father:
 - Determine the possible phenotypes of children they might have

Solutions to Sample Genetics Problems

- This problem involves a trait that exhibits codominance, and the gene is located on the X chromosome in cats; to solve this problem, assume sex determination in cats is the same as for humans

Genetics Key:
 $X^B = \text{black}$, $X^O = \text{orange}$, $Y = \text{no allele for this trait}$
 $X^B X^B = \text{black female}$
 $X^B X^O = \text{calico female}$
 $X^O X^O = \text{orange female}$
 $X^B Y = \text{black male}$
 $X^O Y = \text{orange male}$

	X^B	X^O
X^B	$X^B X^B$	$X^B X^O$
Y	$X^B Y$	$X^O Y$

The Cross:
male = $X^B Y \times$ female = $X^B X^O$

Answers:

 - 50% of the female kittens will be calico
 - 0% of the male kittens will be calico (the two codominant alleles must both be present to produce a calico phenotype; males can only have one allele, as they only have one X chromosome)
- This problem is a basic two-trait, progeny-testing cross as originally described by Mendel

Answers:

 - P generation: either GGWW, ggww **OR** GGww, ggWW
 - Gametes: either GW, gw **OR** Gw, gW
 - F_1 individuals: GgWw

- $F_1 \times F_1$ cross yields nine genotypes in the F_2 individuals
 - $F_1 \times F_1$ cross yields a phenotypic ratio of **9:3:3:1**
- Both fur color and ear length display complete dominance; however, both genes are linked, and thus, when calculating gamete possibilities, be aware that the two genes will NOT assort independently

Genetics Key:
Fur Color: $B = \text{blue}$, $b = \text{red}$
 $BB = \text{blue}$
 $Bb = \text{blue}$
 $bb = \text{red}$

Ear Length: $L = \text{long}$, $l = \text{short}$
 $LL = \text{long}$
 $Ll = \text{long}$
 $ll = \text{short}$

The Cross:
BBLL x bbll → BbLl

- Answers:**
- One phenotype produced: Blue fur, long ears
 - 2 (BL, bl; because the dominant and recessive alleles are on separate chromosomes, respectively)
 - The test cross of BbLl x bbll **without crossing over** yields the following results:

	BL	bl
bl	BbLl	bbll

Two phenotypes produced: (1) Blue fur, long ears; (2) Red fur, short ears

- The test cross of BbLl x bbll **with crossing over** yields the following results*:

	BL	Bl	bL	bl
bl	BbLl	Bbll	bbLl	bbll

Four phenotypes produced: (1) Blue fur, long ears; (2) Blue fur, short ears; (3) Red fur, long ears; (4) Red fur, short ears
[* NOTE: It is possible crossing over would not recombine the dominant and recessive alleles; in such case, only two phenotypes ("c" above) would be produced; however, because crossing over might produce recombinant progeny, the answer reflects the possible gametes and progeny]

- This problem involves two separate inheritance patterns: ABO blood group and diabetes [see **Genetics Key** in solution to question #6]

The Cross:

Stephan = BOdd x Jenna = AODd; Oscar = OOdd (to produce this child's genotype and phenotype, both parents must have "O" alleles for blood type and "d" alleles for diabetes)

	AD	Ad	OD	Od
Bd	ABDd	ABdd	BODd	BOdd
Od	AODd	AOdd	OODd	OOdd

Answers:

- a. & b. $\frac{1}{8}$ offspring will have type O blood and be diabetic (genotype = OOdd)
5. Carly, Cedric and both sets of parents have normal phenotypes (otherwise, they would have stated they have the disease); for Cedric and Carly's father, the genotype/phenotype are: X^HY = **normal male** (it does not matter that Cedric's cousin has the disease); however, Carly's genotype/phenotype can be: X^HX^H or X^HX^h = **normal female**; but because Carly has a brother with hemophilia (genotype/phenotype: X^hY = hemophiliac), their mother would have to contribute a gamete with the hemophilia allele; thus, the mother's genotype is X^HX^h , which is a normal phenotype but carrier genotype; following is a Punnett square with the cross of Carly's parents:

	X^H	X^h
X^H	X^HX^H	X^HX^h
X^h	X^HX^h	X^hX^h

Answer:

- a. Carly's genotype cannot be definitively determined
 Carly's probability of being X^HX^h = 50% (0.5)
 Probability of a daughter with the disease = 0% (0.0)
 0.5 Carly is a carrier
 x $\frac{0.0}{0.0}$ Daughter is diseased
 0.0
 0% overall probability of daughters with the disease
 Probability of a son with the disease = 50% (0.5)
 0.5 Carly is a carrier
 x $\frac{0.5}{0.25}$ Son is diseased
 0.25
 25% overall probability of sons with the disease

6. This problem involves two separate inheritance patterns: Cattle fur color, which exhibits incomplete dominance; and horn development, which exhibits complete dominance

Genetics Key:

Fur Color: W = white, w = red
 WW = white
 Ww = roan
 ww = red

Horn Development: H = hornless
 h = horns present
 HH = hornless
 Hh = hornless
 hh = horns present

The Cross:

The use of the symbol "F₁" traditionally indicates progeny testing, as originally employed by Mendel in his pea plant crosses; this requires that the original parents be pure breeders or homozygous, and that the hornless genotype must be HH , not Hh ; thus, the P generation crosses could be either: $WWHH$ x $wwhh$ OR $WWHh$ x $wwHH$

Answers:

- a. F₁ genotype: $WwHh$
 F₁ x F₁ cross:

	WH	Wh	wH	wh
WH	$WWHH$	$WWHh$	$WwHH$	$WwHh$
Wh	$WWHh$	$WWhh$	$WwHh$	$Wwhh$
wH	$WwHH$	$WwHh$	$wwHH$	$wwHh$
wh	$WwHh$	$Wwhh$	$wwHh$	$wwhh$

- b. 6 out of 16 (38%) would be **roan & hornless**, with genotypes of either $WwHH$ or $WwHh$
- c. 1 out of 16 (6%) would be **white & horned**, with a genotype of $WWhh$
7. The key to solving this problem is that the sister is color-blind, which is the genotype/phenotype: X^cX^c = **color-blind female**; this means that both parents contributed the " X^c " allele; the other sister was normal, which would make her genotype/phenotype: X^CX^c = **normal-vision female**; thus, the mother must have contributed the " X^c " or normal allele

Answer:

- a. Male parent: X^cY = color-blind male
 Female parent: XCX^c = normal-vision female
8. This problem involves a trait involving complete dominance; in this case, the normal or "good" allele is dominant, and the defective allele that causes diabetes is recessive:

Genetics Key:

D = normal, d = diabetes
 DD = normal, Dd = normal, dd = diabetic

The Cross:

Both parents must be heterozygotes:
 Dd x Dd = dd child

Answer:

- a. 25% probability for all subsequent offspring to have the same genotype/phenotype as the first child
9. To solve this problem, and others that are unfamiliar, first create a key for symbols and the type of genetic inheritance pattern; foot growth direction displays complete dominance, while foot size displays incomplete dominance:

Genetics Key:

Foot Growth Direction:
 B = forward, b = backward
 BB = forward
 Bb = forward
 bb = backward

Foot Size:
 L = large or big, l = small or little
 LL = large feet
 Ll = normal feet (incomplete dominance)
 ll = small feet

The Cross:

Male = $bbLL$ x Female = $BbLl$

Answers:

- a. The female can produce two gamete types (BL , Bl)
- | | | |
|-----------|-----------|-----------|
| | BL | Bl |
| bL | $BbLL$ | $BbLl$ |
- b. Two phenotypes produced: (1) forward-pointing, big feet; (2) forward-pointing, normal-sized feet
- c. Changing the original cross to male = $bbLl$ x female = $BbLl$ yields the results shown:
- | | | |
|-----------|-----------|-----------|
| | BL | Bl |
| bL | $BbLL$ | $BbLl$ |
| bl | $BbLl$ | $Bbll$ |
- d. Changing the original cross to male = $bbLL$ x female = $bbLl$ yields the results shown:
- | | | |
|-----------|-----------|-----------|
| | bL | bl |
| bL | $bbLL$ | $bbLl$ |
- e. Changing the original cross to male = $bbLL$ x female = $bbLl$ would yield no offspring with forward-pointing feet

10. This problem involves two separate inheritance patterns in humans: ABO blood group and color-blindness

The Cross: Randy = $AB X^cY^c$; Sheri = $AO X^CX^c$ (this must be her genotype, as her mother contributed an "O" allele for blood type, and her father contributed an " X^c " allele for color-blindness)

	AX^c	AY	BX^c	BY
AX^c	AAX^cX^c	AAX^cY	ABX^cX^c	ABX^cY
AY	AAX^cX^c	AAX^cY	ABX^cX^c	ABX^cY
OX^c	AOX^cX^c	AOX^cY	BOX^cX^c	BOX^cY
OY	AOX^cX^c	AOX^cY	BOX^cX^c	BOX^cY

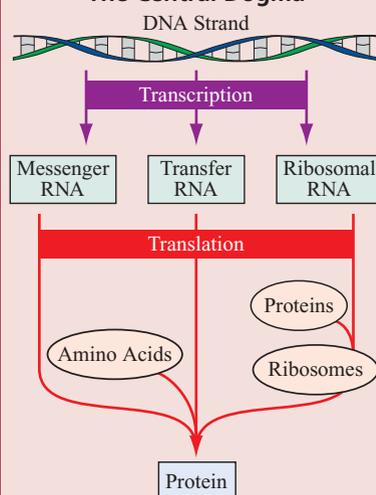
Answer:

- a. Male and female offspring would have every combination of blood type A, B & AB with every combination of vision (i.e., normal vs. color-blindness); based on the known genetics patterns, however, type O blood would not be present in any offspring, regardless of gender

Molecular Genetics**A. The Central Dogma**

1. Soon after the discovery of the structure of DNA, the function of nucleic acids in general was better understood; the following subsection summarizes the major processes involved in **molecular biology**; when relating these processes to inheritance, this is called **molecular genetics**:

- a. **DNA Replication:** New DNA is copied from existing DNA—a process that uses or conserves half the original DNA, while the other half is new (i.e., semiconservative replication)
- b. **DNA Transcription:** Messenger RNA (mRNA) is copied from DNA; intervening sequences (**introns**) are removed and remaining portions (**exons**) are ready to be translated
- c. **DNA Translation:** Proteins are synthesized from mRNA by ribosomes

The Central Dogma

2. **Retroviruses:** Group of RNA viruses possessing a special enzyme, **reverse transcriptase**, that allows the viral genome to go in the reverse direction of the information flow of the central dogma; this allows the viral RNA to be converted into DNA, thereby altering the host cell's genome

a. DNA ← Viral DNA**B. Mutations**

1. Any random, permanent change in the DNA molecule; many are harmful, some have no effect, and a few actually benefit the organism; nature selects those mutations that are beneficial or adaptive in organisms to help shape the course of evolution
- a. **Point Mutation:** Change in one nucleotide base pair
- b. **Chromosomal Mutation:** Change in chromosome number or structure

C. Genomics

1. Study of the entire genome of species
- a. **Structural Genomics:** Gene mapping and sequencing
- b. **Functional Genomics:** Studying how gene sequences operate, including **proteomics** (study of functioning of proteins coded by genes)
- c. **Comparative Genomics:** Analysis of gene sequences of different species

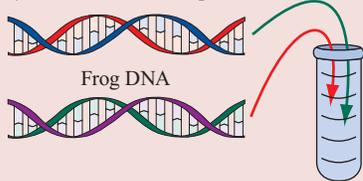
D. Developmental Genetics

1. Advances in molecular biology have revolutionized the study of developmental biology; for example, studies on the fruit fly *Drosophila* have illustrated fundamental processes in **cellular differentiation**

- Homeotic Genes:** Sets of genes that control **basic body patterns** in organisms, including members of the fungal, plant and animal kingdoms
- Homeobox:** Specific nucleotide sequences (composed of about 180 base pairs) typically associated with homeotic genes
- Hox Genes:** Subset of homeobox genes that are found in many different animals and are highly conserved (i.e., basically the same), indicating that, evolutionarily, they arose very early in the development of life on Earth

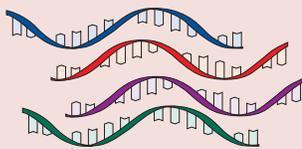
Homeobox Hox Genes of Animals Are Highly Conserved

Fly homeobox DNA sequence



Fly homeobox DNA sequence is mixed with Frog DNA

Heat



Strands separate

Cool



Fly homeobox DNA binds to frog DNA, confirming sequence similarity

E. Cancer Genetics

- Cancers frequently involve three types of mutated genes:
 - Oncogenes:** Stimulate abnormal cell growth and division, which can lead to malignant tumors; originate from normal genes, called **proto-oncogenes**,

which regulate the cell cycle; viruses can also transmit oncogenes to host cells

- Tumor-Suppressor Genes:** Normally prevent the uncontrolled growth and division of cells and tissues; *mutations may deactivate these genes*, which can lead to cancers
- Mutator Genes:** Can increase the mutation rates of other genes, thereby increasing the chance of cancerous tissues developing

F. Non-Mendelian Inheritance

1. Mitochondrial & Chloroplast Genomes

- The **Endosymbiotic Hypothesis** suggests that mitochondria and chloroplasts are derived evolutionarily from prokaryotes; the strongest evidence for this hypothesis is that both organelles have their own set of DNA: **mtDNA** in mitochondria and **cpDNA** in chloroplasts
- Genes on these non-nuclear sources of DNA are called **extranuclear genes**; they have “non-Mendelian” inheritance patterns, because they **do not engage** in meiotic segregation and re-assortment processes

c. **Uniparallel Inheritance:** Offspring (male and female) have *characteristics of just one parent*

d. **Maternal Inheritance:** In most sexually reproducing, eukaryotic organisms, the haploid genome and cytoplasm with organelles from the female gamete are passed to the zygote, while the male gamete essentially contributes only a haploid genome; very little cytoplasm is transferred to the zygote; thus, *traits associated with mtDNA are inherited from the maternal parent*

2. **Infectious Heredity:** Symbiotic bacteria and viruses that are transmitted in cytoplasm

G. Molecular Evolution

1. Although morphological characteristics (visible phenotypes) have been the traditional focus of evolutionary biology, emergent molecular techniques have allowed the study of the evolution of DNA and protein sequences

2. **Molecular Phylogenies:** Phylogenetic trees, illustrating evolutionary relatedness of all species, have been revised and updated using modern molecular techniques; one major development was dividing life into three large groups or domains: **Bacteria, Archaea and Eukarya**

Population & Evolutionary Genetics

Genes in populations versus individuals

- Populations evolve just as species do
- Genotype:** Genetic composition of an individual
- Gene Pool:** Genetic composition of a population of individuals; that is, all alleles for all genes in a population
- Evolution involves changes in gene pools over time; to understand changes in gene pools as populations evolve, an understanding of non-evolving populations is necessary

The Hardy-Weinberg Law

A. Both allelic frequencies and genotypic ratios (i.e., gene pools) remain constant from generation to generation in sexually reproducing populations, if the following conditions of equilibrium exist:

- Mutations do not occur
- No net movement of individuals out of or into a population occurs
- All offspring produced have the same chances for survival, and mating is random; that is, no natural selection occurs
- The population is large so that chance would not alter frequencies of alleles

B. Algebraic equivalent of the Hardy-Weinberg Law:

- $p^2 + 2pq + q^2 = 1$, where:
 - p = frequency of dominant allele
 - q = frequency of recessive allele
 - p^2 = AA genotype
 - $2pq$ = Aa genotype
 - q^2 = aa genotype

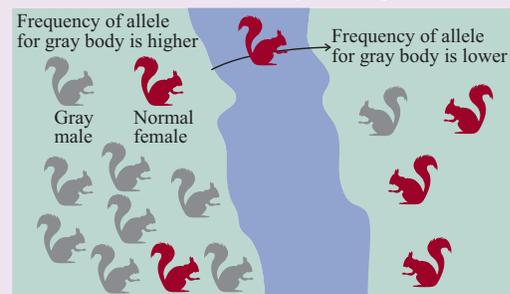
C. Example:

- If, in a group of six individuals, there are nine dominant (A) alleles and three recessive (a) alleles, then $p = \frac{9}{12}$ or 0.75 and $q = \frac{3}{12}$ or 0.25; a total of 12 gametes will be produced, nine of which will have the dominant allele and three with the recessive allele
- The algebraic equation above can be used to predict the ratios of the three possible genotypes as a result of fertilizations:
 - Frequency of AA genotypes is p^2 or $(0.75)^2 = 0.56$
 - Frequency of Aa genotypes is $2pq$ or $2(0.75)(0.25) = 0.38$
 - Frequency of aa genotypes is q^2 or $(0.25)^2 = 0.06$
- The frequencies of dominant and recessive alleles are still the same—the specific alleles have been redistributed

Hardy-Weinberg & Natural Populations

- Few (if any) populations are in equilibrium; therefore, changes in allele frequencies and, thus, gene pools do occur in natural populations
- The Hardy-Weinberg Law helps to identify the mechanisms of these evolutionary changes by predicting that one or more of the four conditions required are not met; that is:
 - Mutations occur
 - Individuals leave and enter populations
 - Non-random mating and natural selection occur
 - Small populations exist

Allele Frequency Changes



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