Chapter 17

Patterns and Processes in Inheritance

Chapter Concepts

- Describe the evidence for segregation and the independent assortment of alleles
- Compare ratios and probabilities of genotypes and phenotypes for traits involving a single gene
- Demonstrate the inheritance of a trait controlled by a single gene
- Interpret patterns and trends in data that form monohybrid and dihybrid inheritance

Chapter Concepts

- Explain inheritance patterns for genes on the same chromosome
- Analyze crossing over data and create a chromosome map for genes on a single chromosome
- Describe inheritance patterns for sex-linked genes
- Compare ratios and probabilities of genotypes and phenotypes for multiple alleles and for polygenic traits

Chapter Concepts

- Investigate the influence of environmental variables on the expression of genetic information in an individual
- Describe ways in which plant and animal breeding programs use genetic research
- Draw and interpret pedigree charts that show the inheritance of traits in humans

Chapter Concepts

- Investigate the inheritance of human traits
- Assess the role of genetic counselling and technology in issues that involve society
- Evaluate some of the social, ethical, and economic considerations that are involved in the application of genetic research

17.1 - Early Genetics

- Even early cultures and civilizations had a rudimentary knowledge of genetics
- They understood that the breeding of individuals with desirable characteristics often produced offspring with these characteristics as well

Gregor Mendel & Genetics

- Mendel was a monk who is considered the father of genetics
- Using pea plants he studied how certain traits are passed from generation to generation
- he found that some traits were recessive (only showed up in purebred strains) while others were dominant

http://web.educastur.princast.es

- He determined that **dominant** genes would be expressed whenever they were present, and the **recessive** genes are only expressed if there are no other dominant genes present
- He also developed 3 laws of heredity:

Mendel's Laws of Genetics

1. Inherited characteristics are controlled by genes that occur in pairs. In cross-fertilization, each parent contributes one gene.
2. One gene can mask another. The dominant gene is expressed using an upper case letter while the recessive uses a lower-case letter.
3. Pairs of genes separate when gametes form. This is known as the law of **segregation.**

Terms Used in Genetics

- **Genotype**:
- **Phenotype:**
- **Homozygous**:
- **Heterozygous**:

A Few More Terms…

- **Alleles**:
- **Purebred:**
- **Hybrid:**

Single-Trait Inheritance: Punnett Squares

- The Punnett square can be used to determine the inheritance of single traits
- The border spaces of the square are used for the gametes produced by the parents
- The inner spaces represent the possible genotypes for the offspring

Example: Crossing of Round & Wrinkled Peas

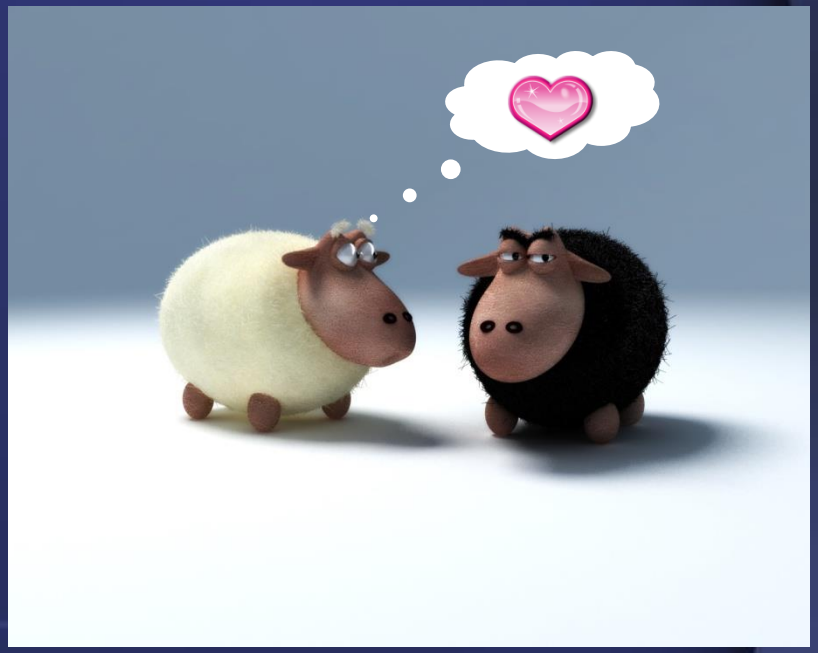
- In this case, a wrinkled pea (recessive) is crossed with a heterozygous round pea

Test Crosses

- Often you know the genotypes of the offspring, but not of the parents
- Therefore, a Punnett square can be used to determine the genotype of a parent that has a dominant phenotype
- In these test crosses, the other parent used must be one that is homozygous recessive

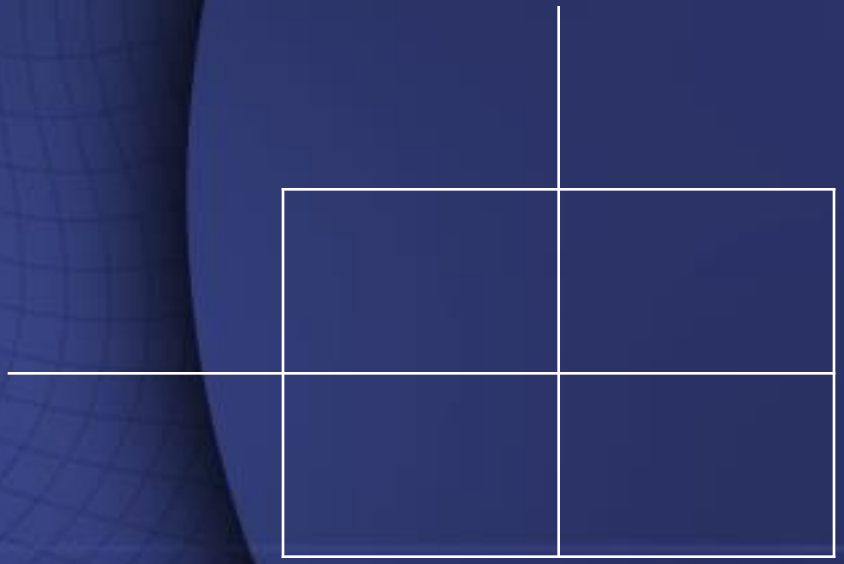
Example: Test Cross of White & Black Sheep

- In this case, black wool colour is a recessive trait
- To determine the genotype of a white sheep, a test cross is performed
- The phenotypic ratio of the offspring will determine the genotype of the white sheep



Possible Results

- Case 1: White sheep is heterozygous



- Case 2: White sheep is homozygous

Dihybrid Crosses

- All organisms have multiple traits
- We can use a modified Punnett square to determine the expected genotypes and phenotypes for crosses involving two traits
- To keep these simple, we limit ourselves to two traits at a time
- We must also assume that the law of independent segregation also holds true

Example: Cross-bred Aliens

- Ex: An alien species known as Biothirtians, are captured and bred by the government in area 51A. The scientists are studying two genes at a time. The first trait is colour. The blue skin colour of these aliens seems to be dominant to orange skin colour. The second trait is presence of a long or short tail. Short tails are dominant to long tails. A homozygous blue alien with a long tail is mated with an orange alien with a homozygous short tail.

http://www.turbosquid.com

a) Determine the genotypes and phenotypes of the F_1 generation

	Blue, long-tailed alien				
Orange, short-tailed alien	Gametes				

b) Based on the F_1 generation, determine the possible phenotypes of the F_2 generation

	Blue, short-tailed alien				
Blue, short-tailed alien	Gametes				

Incomplete Dominance

- When incomplete dominance occurs, the resulting phenotypes are a mixture of the phenotypes of the parents
- For instance, if a red snapdragon (C^RC^R) is crossed with a white snapdragon (C^WC^W), the resulting offspring are pink (C^RC^W)

Incomplete Dominance in Humans

- Two genetic conditions in humans show incomplete dominance:

1. Sickle-cell anemia (Hb^S). A person with a heterozygous genotypes (Hb^AHb^S) have the sickle-cell trait, but much fewer of their cells are sickle-shaped than homozygous individuals. This may provide a *heterozygote advantage*.

2. Familial hypercholesterolemia is a genetic condition where tissues cannot remove LDL from the blood. People who are homozygous have six times the normal amount of LDL in the blood. Some may even have heart attacks by age 2!

   Those who are heterozygous only have about twice the normal level of cholesterol.

Co-Dominance

- In co-dominance, both genes are expressed equally
- For instance, the blue roan horse has both a black and white allele
- This actually causes some hairs to be black, while others are white

http://globalhorseculture.typepad.com

Chromosome Theory

- Despite the work that he did, Mendel did not try to explain the material in the cell that carried the traits
- In 1902, Walter Sutton examined synapsis (the segregation of homologous chromosomes) in sperm
- He realized that the pattern of chromosome distribution matched Mendel's law of segregation
- He published a paper that proposed that genes were carried on chromosomes

Chromosomal Theory

- the chromosomal theory of inheritance can be summarized as four points:

1. Law of Dominance

2.Law of Segregation

Chromosomal Theory (Cont.)

3. Law of independent Assortment

4. Genes exist at specific locations (loci) on chromosomes within the nucleus

17.2 – Extending Mendel's Laws

- Thomas Hunt Morgan started to study fruit fly genetics shortly after Sutton proposed his chromosomal theory
- Working with these fruit flies, he made some significant discoveries, such as sex-linked traits and linked genes

Gene Linkage and Crossing–Over

- in Morgan's work with fruit flies, he found that some combinations of traits did not distribute themselves as expected by Mendelian genetics
- he noted that some combinations resulted in phenotypic ratios different from the expected 9:3:3:1 produced by dihybrid crosses
- he suggested that genes on the same chromosomes will not sort independently of each other, and tend to be transmitted together (these are **linked genes**)

Gene Linkage (Cont.)

- however, these genes can be redistributed during meiosis by crossing-over of chromosomes
- this means that a single chromosome can change as it passes between generations
- crossover frequency can be expressed as a percentage:

(# of recombinations / # of offspring) X 100%

Mapping Chromosomes

- a recessive characteristic can be used as a **gene marker** which identifies other characteristics on the same chromosome (if they are on the same chromosome, they will appear together more often)
- however, when crossing-over occurs, it can alter gene linkages

- it was found, through numerous observations and experiments, that genes near each other on a chromosome almost always end up together after crossing-over
- likewise, if two genes are far apart on a chromosome, they are more likely to be affected by crossing-over
- this means that genes with a cross-over value of 1% are much closer together than ones with a value of 12%

Gene Maps Using Crossover Frequencies

- this crossover frequencies are related to **map distance**, or the distance between genes on a chromosome
- 1% = 1 map distance unit
- the frequency of crossovers can be used to determine gene maps

Chromosome Map Example

- Ex: The crossover frequency between genes A and B is 12%, B and C is 7% and A and C is 5%. Arrange the genes in order that they appear on the chromosome.

Jumping Genes

- Some genes can shift throughout a chromosome
- These “jumping genes” were first suggested by Barbara McClintock in the 1940s
- These transposable genes are known as **transposons**

http://www.amphilsoc.org

Sex-Linked Traits and Sexual Determination

- Thomas Hunt Morgan, determined that one pair of chromosomes, the **sex chromosomes**, are not always identical
- by studying *Drosophila*, he determined that males have one X and one Y chromosome, while females had two X chromosomes

Ex: Eye Colour in Drosophila

- A red-eyed female (X^RX^R) is bred together with a white-eyed male (X^rY). White eye colour is recessive. Determine the genotypes and phenotypes of the possible offspring:

http://www.biology.ualberta.ca

The F_2 Generation

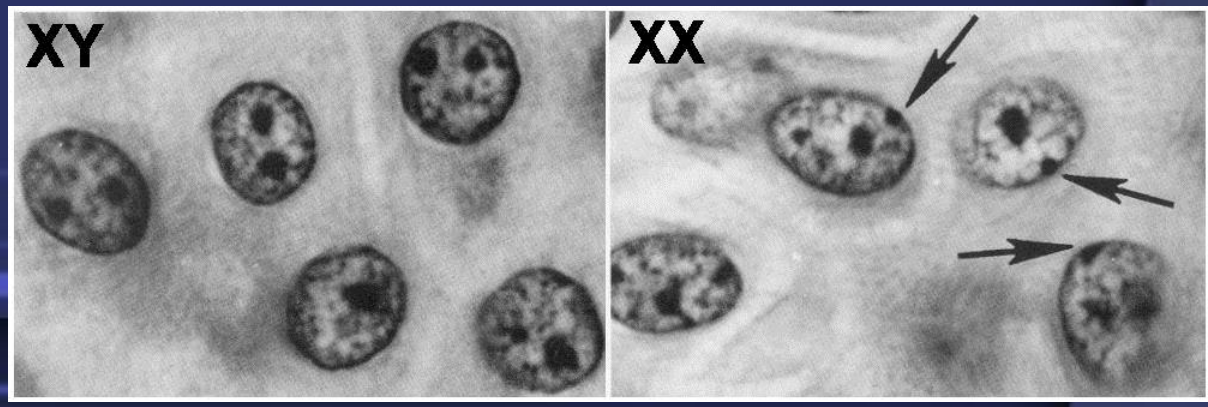
- What are the results of crossing one F_1 male and one F_1 female from the previous example?

Sex Determination

- the male and female chromosomes have homologous regions (identical regions) and differential regions
- often sex-linked traits are carried on the differential region of the X chromosomes

Determining Gender – Barr Bodies

- female cells can often be identified by a black spot called a **Barr body,** which is seen in somatic cells during interphase
- Mary Lyon, a British scientist, proposed that these bodies are X chromosomes that are inactive (which turned out to be true – some genes are turned “on” or “off” in the cells)



http://www.mun.ca

Barr Bodies – A Clue to How Genes Work?

- this is how body cells can carry out very different functions despite having the same DNA
- For example, calico cats carry one X chromosome that carries an orange allele while the other X chromosome carries the black allele for coat colour
- In these cats, the orange fur patches has the orange X chromosome active, while the black fur patches have the black X chromosome active

Gender Verification

- females who have Turner's syndrome (a single X chromosome) will not exhibit Barr bodies
- if these women were tested for Barr bodies to confirm gender (such as in the Olympics) they would test negative
- in other cases, such as testicular feminization syndrome, XY individuals appear to be females despite the presence of Y chromosomes
- this is because the individual does not react to injections of male sex hormones & therefore do not show the muscle development of males

Multiple Alleles

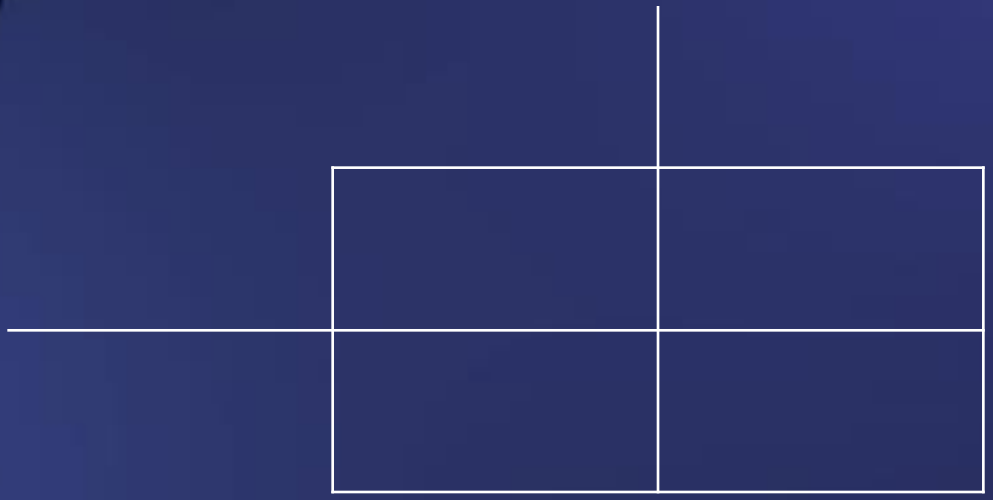
- Often there are more than two alleles for a gene
- As a result, a number of possible different combinations are possible
- Some multiple alleles show dominance in a hierarchy

Ex: Eye Colour in Drosophila

Phenotype	Genotype	Dominant to
Wild type	E^1E^1, E^1E^2, E^1E^3, E^1E^4	Apricot, honey, white
Apricot	E^2E^2, E^2E^3, E^2E^4	Honey, white
Honey	E^3E^3, E^3E^4	White
White	E^4E^4	

A pair of drosophila (wild type E^1E^4 and apricot E^2E^3) are crossed. What are the phenotypes and genotypes that are produced?

Answer:



Example: Human Blood Types

- Human blood types show a type of inheritance that combines multiple alleles and codominance
- In this case, the genes for type A blood and type B blood are equally dominant over the gene for type O blood

Phenotype	Genotype(s)
A	$I^A I^A$, $I^A i$
B	$I^B I^B$, $I^B i$
AB	$I^A I^B$
O	i i

Ex: Blood Types

- A woman who has type A blood (heterozygous) has children with a man with type B blood (heterozygous)
- Determine the possible genotypes and phenotypes of the offspring

Polygenic Inheritance

- Some traits involve more than one gene
- A number of these polygenic traits are continuous (they have a range of possible forms of a trait)
- For instance, the length of an ear of corn is polygenic

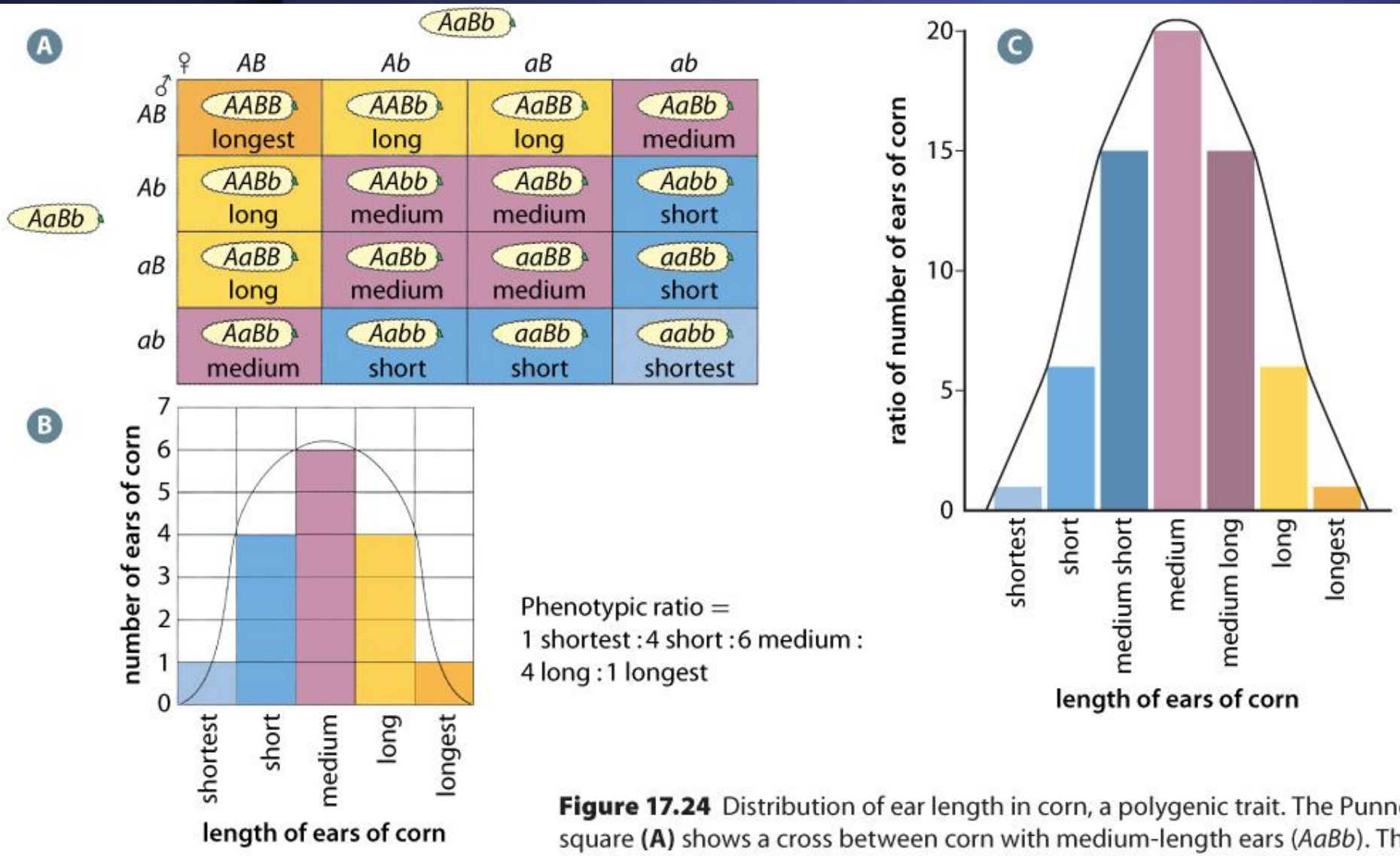
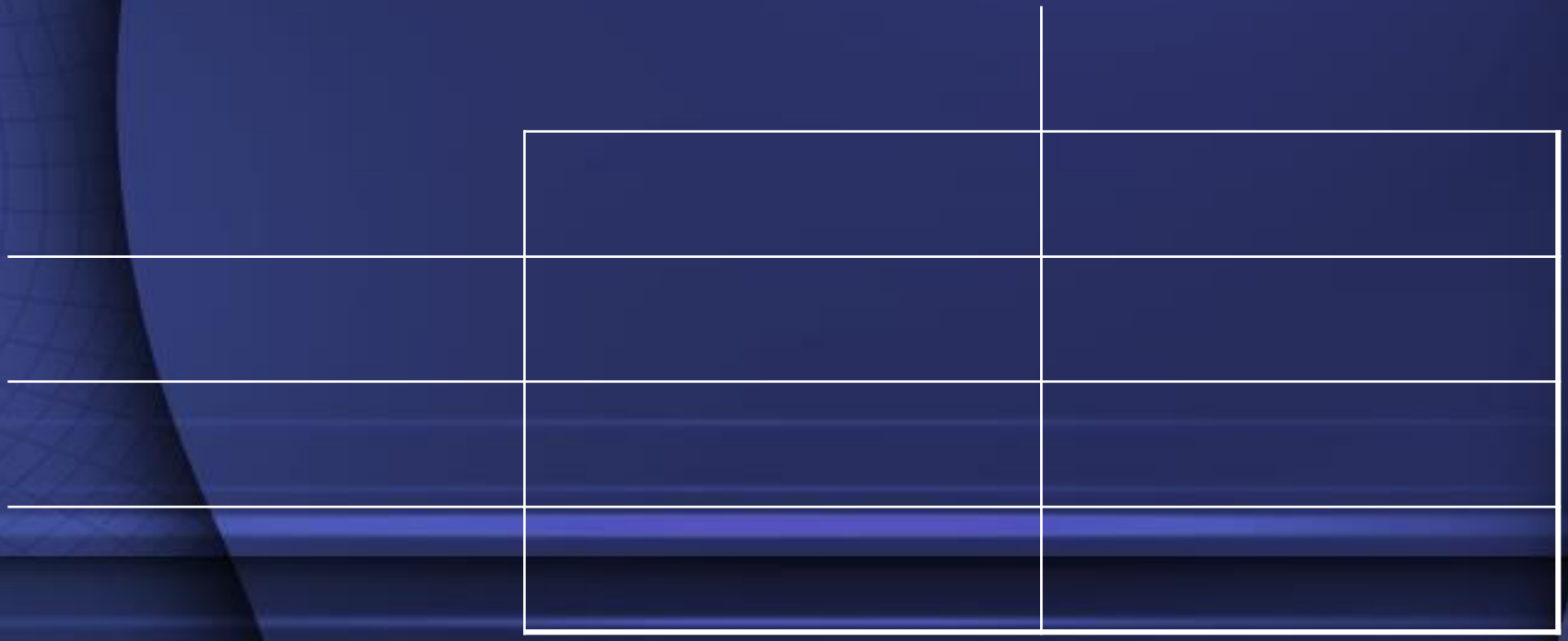


Figure 17.24 Distribution of ear length in corn, a polygenic trait. The Punnett square **(A)** shows a cross between corn with medium-length ears (*AaBb*). The resulting phenotype ratio of 1:4:6:4:1 is graphed in **(B)**. In graph **(C)**, three genes control ear length, resulting in a more gradual-length distribution curve.

Ex: Coat Colour in Dogs

- In dogs, the allele (B) produces a black coat while (b) produces a brown coat. However, another allele, (W) prevents the formation of pigment, while the recessive allele (w) does not prevent colour. What are the expected results of a cross between WwBb and wwBb?

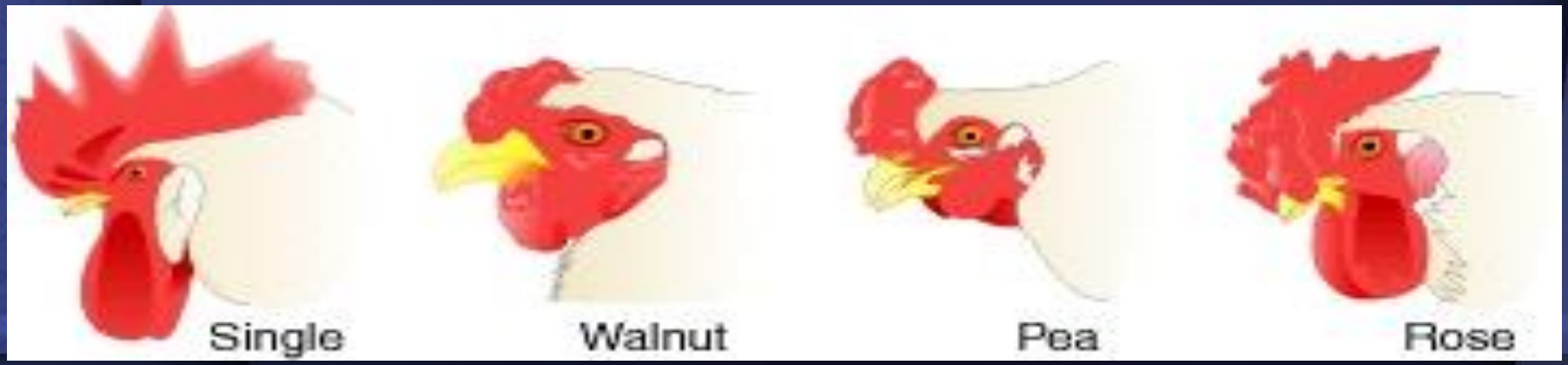


Gene Interactions

- Sometimes the presence of one gene will interfere with the expression of another
- These genes are known as **epistatic** genes
- Other sets of genes work together and are complementary

Ex: Complementary Genes

- The shape of the combs of chickens depends on two different genes. The R allele forms a rose comb, while the P allele on a different gene forms a pea comb. If R and P are both present, they form a walnut comb. If r and p are present, a single comb is produced
- A rose comb (Rrpp) and a pea comb (rrPp) are crossed to produce the F_1 generation



http://www.ncbi.nlm.nih.gov/books/NBK21430/

- Results of the F_1 generation offspring

	Gametes				

Genes & Proteins

- Ultimately, the purpose of a gene is to code for a protein
- The **Central Dogma** of molecular biology is “DNA to RNA to protein”
- In other words, each gene codes for a single protein
- Therefore, DNA can regulate chemical reactions in a cell by controlling the production of enzymes

- Therefore, if a particular compound is produced through a metabolic pathway that uses several enzymes, it is essential that all of the enzymes are present
- If the gene for one of these enzymes is inactivated or not present, then the final product cannot be produced

Pleiotropic Genes

- Pleiotropic genes are single genes that may be responsible for many characteristics
- For instance, a single gene for sickle-cell anemia can produce a number of symptoms
- The sickle shape of the cell prevents it from entering capillaries, even though it still can carry oxygen

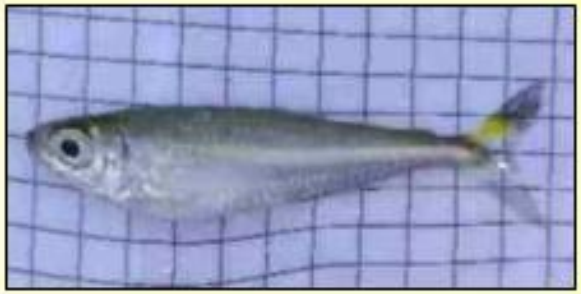
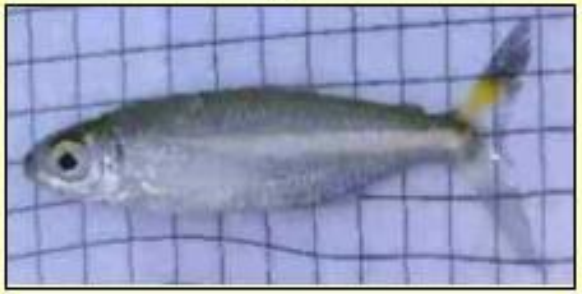
http://medicine.osu.edu

Environment & Phenotype

- The environment is also responsible for the presentation of a particular phenotype
- Often it is difficult to determine whether a phenotype is completely due to the genes that are present, solely due to the environment, or a combination of both

Phenotypic Plasticity

- Some organisms will have physical features that change based on their environment, even if they have the same genes

Species	Channel	Lagoon
Bryconops caudomaculatus		
Biotodoma wavrini		

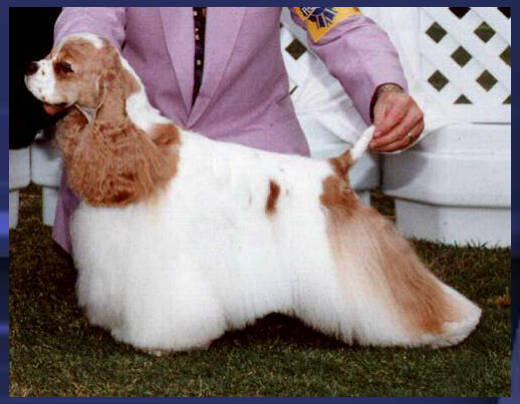
http://wfsc.tamu.edu

17.3 – Genetics & Society

- Humans have been breeding plants and animals to accentuate particular traits for thousands of years
- For instance, canola has been bred since the 1970s to reduce the levels of erucic acid and glucosinolates to make it fit for human consumption

Breeding Animals

- Many animals have been bred for specific traits
- For instance, cattle and pigs are being bred to produce leaner meat, chickens are bred to produce lower-cholesterol eggs, and many varieties of pets are produced through selective breeding

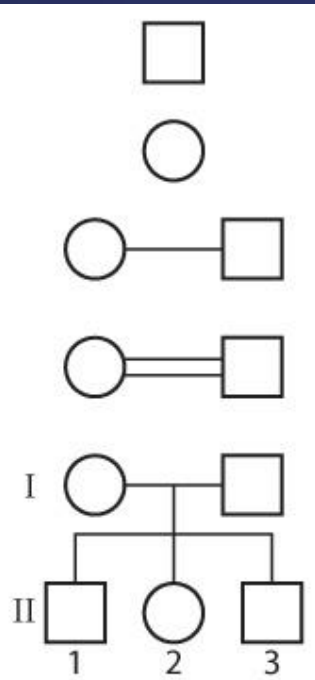


Human Genetics

- Unlike animals, it is difficult to perform experimental crosses with humans
- Therefore, it is difficult to perform analysis of the inheritance of traits due to the low number of offspring
- Instead, we use family history to study the inheritance of traits
- This is done using a pedigree

Pedigree Symbols

Figure 17.28 To make comparisons between different pedigrees as easy as possible, geneticists use several symbols to prepare a pedigree.



male

female

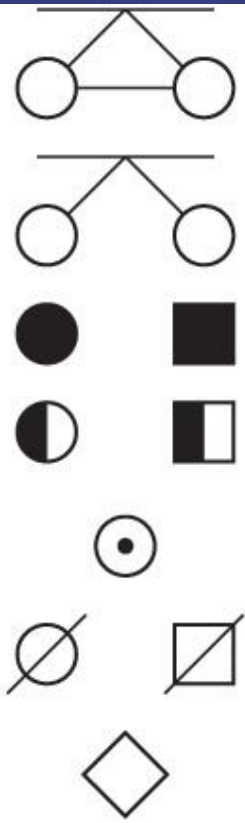
mating

mating between close relatives

Roman numerals symbolize generations.

Arabic numbers symbolize individuals within a given generation.

Birth order, within each group of offspring, is drawn left to right, first-born to last-born.



identical twins

non-identical twins

affected individuals

known heterozygotes for autosomal recessive trait

known carrier of X-linked recessive trait

deceased individuals

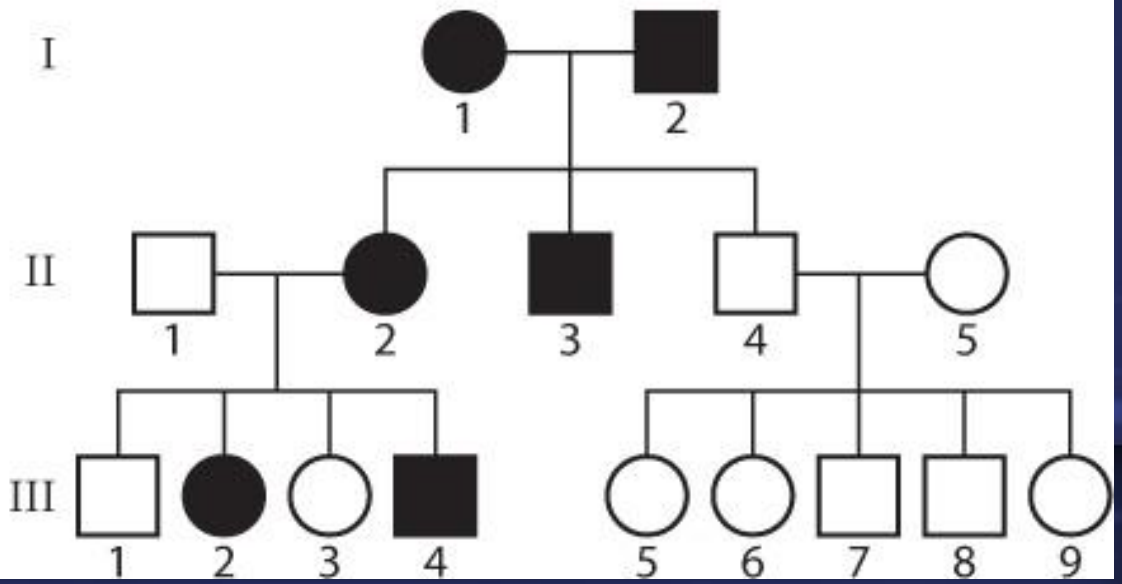
sex unknown

Autosomal Dominance

- These refer to traits that are found on the autosomes and show dominant-recessive inheritance patterns
- If a trait is dominant, only one parent needs to pass on the allele to the offspring for the trait to be expressed

Dominant Autosomal Trait - Polydactyly

Figure 17.29 This pedigree shows the inheritance of polydactyly—an autosomal dominant trait. Notice that heterozygotes are affected by the trait, and that an affected child must have at least one affected parent. How do you know that individual II 2 is heterozygous?



Autosomal Recessive Traits

- Traits that are autosomal recessive (such as phenylketonuria – PKU or cystic fibrosis) must have a gene passed on from each parent
- Often, a distinct pattern where a trait "skips" a generation appears with this type of trait

Autosomal Recessive Trait - PKU

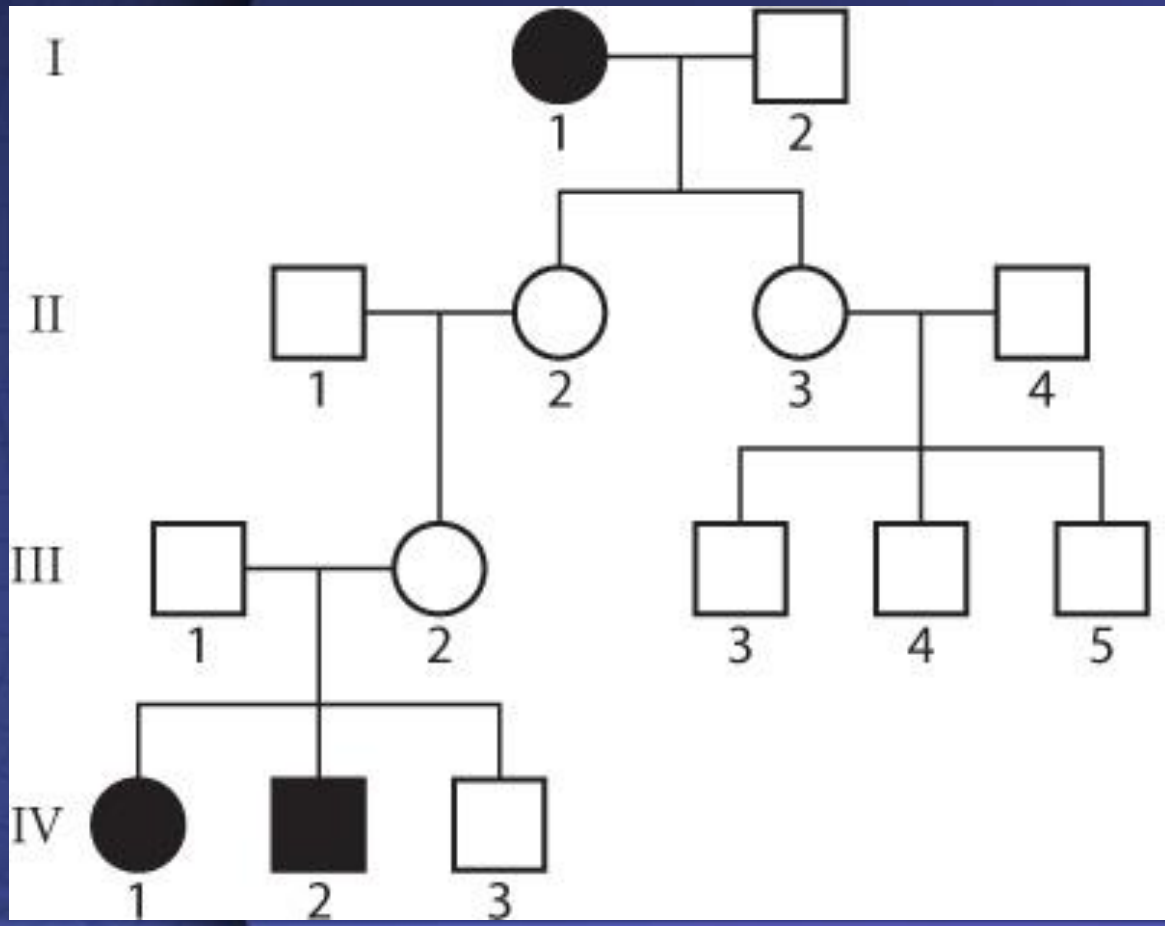


Figure 17.30 A pedigree showing the inheritance of PKU, an autosomal recessive trait. Notice that affected children may have unaffected parents, and that heterozygotes are not affected by the trait. How do you know that individual III 1 is heterozygous?

Sex-Linked Traits

- Sex-linked traits occur more often in males than females (recall how sex-linked traits work)
- Women who do not exhibit the trait pass the trait on to their male children in most cases
- Hemophilia, colour blindness, and Duchenne muscular dystrophy are all sex-linked traits

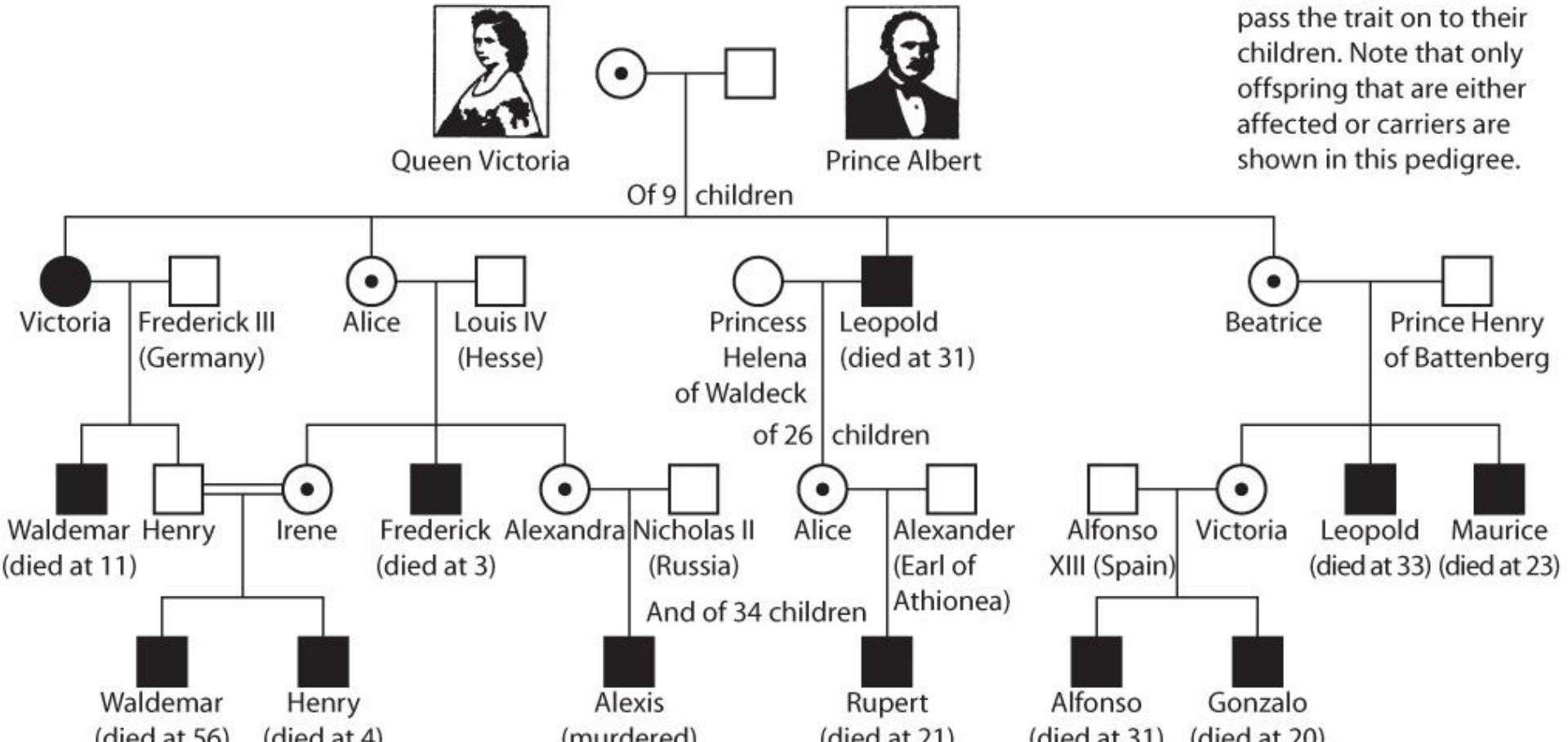


Figure 17.31 Pedigree showing the inheritance of hemophilia, an X-linked recessive trait, in the European Royal families. Males are much more likely to have hemophilia than females. Females who are heterozygous for the condition (carriers) can pass the trait on to their children. Note that only offspring that are either affected or carriers are shown in this pedigree.

Human Genetic Analysis

- Since the early 1900s, it was understood that a number of disorders and illnesses were inherited
- We can now use chromosome mapping to help to determine who are at risk of developing a particular inherited condition or of passing the trait on to their children

Genetic Counsellors

- Genetic counsellors often speak with people who either have a particular genetic condition or are worried about passing the condition on to their children
- They can also explain the symptoms of genetic conditions and the available treatments, or give emotional support