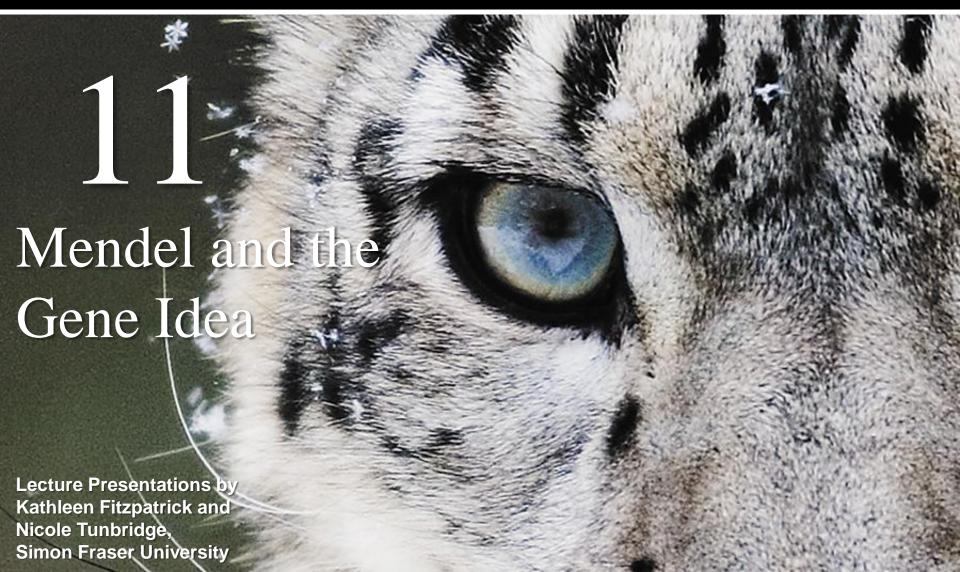
CAMPBELL BIOLOGY IN FOCUS

URRY • CAIN • WASSERMAN • MINORSKY • REECE



Overview: Drawing from the Deck of Genes

- What genetic principles account for the passing of traits from parents to offspring?
- The "blending" hypothesis is the idea that genetic material from the two parents blends together (the way blue and yellow paint blend to make green)

- The "particulate" hypothesis is the idea that parents pass on discrete heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas

Figure 11.1



© 2016 Pearson Education, Inc.

Concept 11.1: Mendel used the scientific approach to identify two laws of inheritance

 Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

Mendel's Experimental, Quantitative Approach

- Mendel probably chose to work with peas because
 - There are many varieties with distinct heritable features, or characters (such as flower color); character variants (such as purple or white flowers) are called traits
 - He could control mating between plants

Technique 0 **Parental** generation (P) Stamens Carpel **Results** 6 First filial generation offspring (F₁)

- Mendel chose to track only characters that occurred in two distinct alternative forms
- He also used varieties that were true-breeding (plants that produce offspring of the same variety when they self-pollinate)

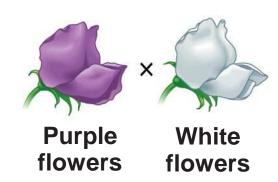
- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called hybridization
- The true-breeding parents are the P generation
- The hybrid offspring of the P generation are called the F₁ generation
- When F₁ individuals self-pollinate or cross-pollinate with other F₁ hybrids, the F₂ generation is produced

The Law of Segregation

- When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F₁ hybrids were purple
- When Mendel crossed the F₁ hybrids, many of the F₂ plants had purple flowers, but some had white
- Mendel discovered a ratio of about three to one, purple to white flowers, in the F₂ generation

Experiment

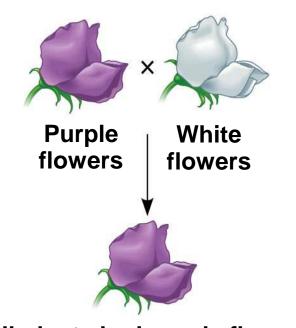
P Generation (true-breeding parents)



Experiment

P Generation (true-breeding parents)

F₁ Generation (hybrids)



All plants had purple flowers

Experiment P Generation X (true-breeding parents) **Purple** White **flowers** flowers **F**₁ Generation (hybrids) All plants had purple flowers **Self- or cross-pollination Results** F₂ Generation 705 purple-flowered 224 white-flowered plants plants

- Mendel reasoned that in the F₁ plants, the heritable factor for white flowers was hidden or masked in the presence of the purple-flower factor
- He called the purple flower color a dominant trait and the white flower color a recessive trait
- The factor for white flowers was not diluted or destroyed because it reappeared in the F₂ generation

- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits
- What Mendel called a "heritable factor" is what we now call a gene

Table 11.1 The Results of Mendel's F ₁ Crosses for Seven Characters in Pea Plants						
Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant: Recessive	Ratio	
Flower color	Purple	×	White	705:224	3.15:1	
Seed color	Yellow	×	Green	6,022:2,001	3.01:1	
Seed shape	Round	×	Wrinkled	5,474:1,850	2.96:1	
Pod shape	Inflated	×	Constricted	882:299	2.95:1	
Pod color	Green	×	Yellow	428:152	2.82:1	
Flower position	Axial	×	Terminal	651:207	3.14:1	
Stem length	Tall	×	Dwarf	787:277	2.84:1	

Table 11.1 The Results of Mendel's F₁ Crosses for Seven Characters in Pea Plants

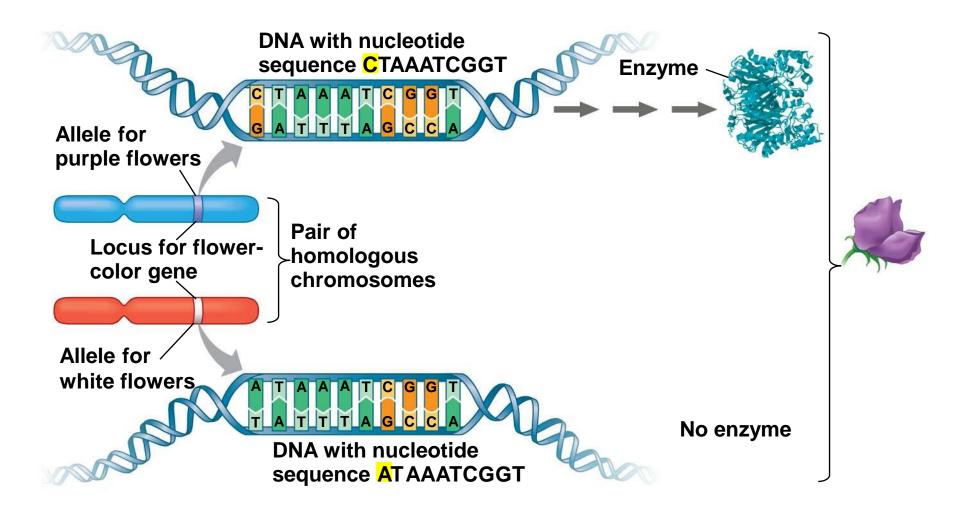
Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant: Recessive	Ratio
Flower color	Purple	×	White	705:224	3.15:1
Seed color	Yellow	×	Green	6,022:2,001	3.01:1
Seed shape	Round	×	Wrinkled	5,474:1,850	2.96:1
Pod shape	Inflated	×	Constricted	882:299	2.95:1

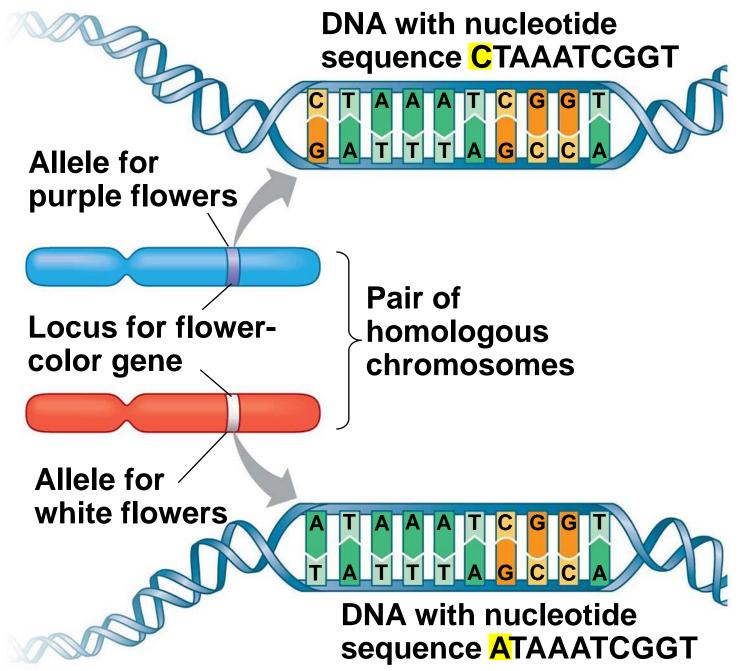
Table 11.1 The Results of Mendel's F ₁ Crosses for Seven Characters in Pea Plants						
Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant: Recessive	Ratio	
Pod color	Green	×	Yellow	428:152	2.82:1	
Flower position	Axial	×	Terminal	651:207	3.14:1	
Stem length	Tall	×	Dwarf	787:277	2.84:1	

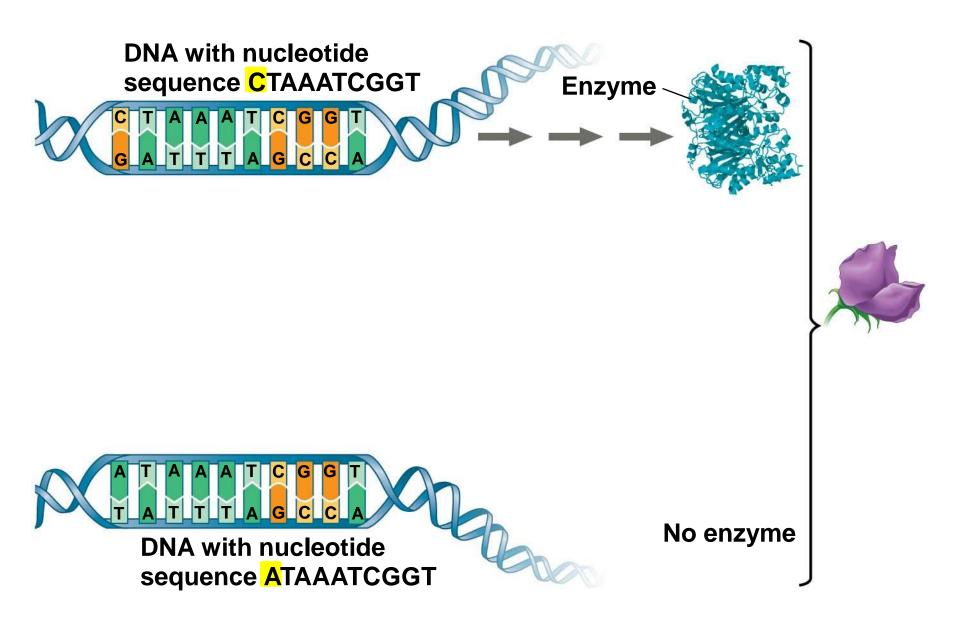
Mendel's Model

- Mendel developed a model to explain the 3:1 inheritance pattern he observed in F₂ offspring
- Four related concepts make up this model

- First, alternative versions of genes account for variations in inherited characters
- For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers
- These alternative versions of a gene are now called alleles
- Each gene resides at a specific locus on a specific chromosome







- Second, for each character, an organism inherits two alleles, one from each parent
- Mendel made this deduction without knowing about the existence of chromosomes
- Two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel's P generation
- Alternatively, the two alleles at a locus may differ, as in the F₁ hybrids

- Third, if the two alleles at a locus differ, then one (the dominant allele) determines the organism's appearance, and the other (the recessive allele) has no noticeable effect on appearance
- In the flower-color example, the F₁ plants had purple flowers because the allele for that trait is dominant

- Fourth (the law of segregation), the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes
- Thus, an egg or a sperm gets only one of the two alleles that are present in the organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis

- Mendel's segregation model accounts for the 3:1 ratio he observed in the F₂ generation of his crosses
- Possible combinations of sperm and egg can be shown using a **Punnett square** to predict the results of a genetic cross between individuals of known genetic makeup
- A capital letter represents a dominant allele, and a lowercase letter represents a recessive allele
- For example, P is the purple-flower allele and p is the white-flower allele

P Generation





Appearance: Genetic makeup:

PP

Purple flowers White flowers pp

Gametes:

P

P Generation







Appearance: Purp Genetic makeup: Gametes:

Purple flowers *PP*

White flowers *pp*

p

F₁ Generation

Purple flowers

Appearance: Genetic makeup:

Pp 1/2 **P**

Gametes:

1/2 (P)

P Generation

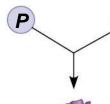






White flowers Appearance: Purple flowers Genetic makeup: PP pp p

Gametes:



F₁ Generation

Purple flowers Appearance: Genetic makeup:

Gametes:



Pp

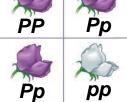
Sperm from

F₂ Generation

 F_1 (*Pp*) plant

Eggs from F_1 (*Pp*) plant

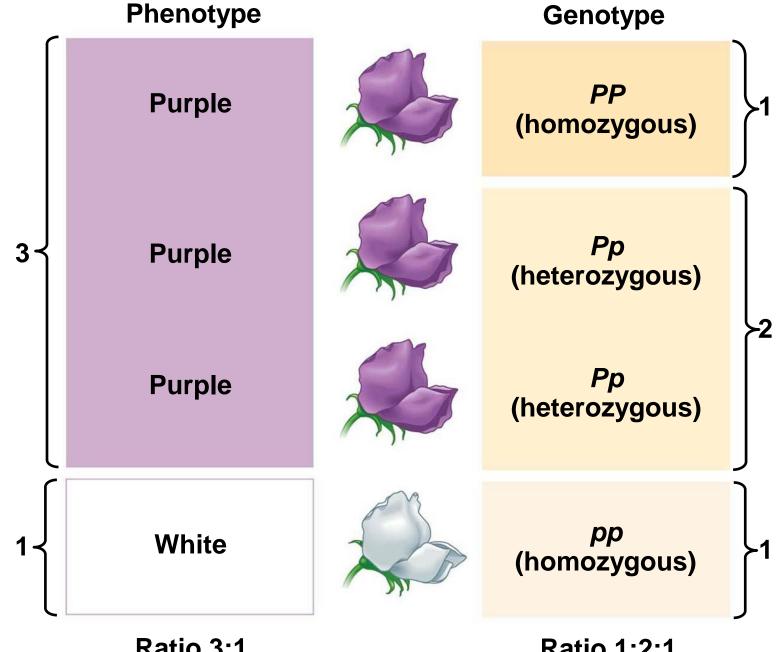
P



Useful Genetic Vocabulary

- An organism with two identical alleles for a character is said to be homozygous for the gene controlling that character
- An organism that has two different alleles for a gene is said to be heterozygous for the gene controlling that character
- Unlike homozygotes, heterozygotes are not truebreeding

- Because of the effects of dominant and recessive alleles, an organism's traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism's phenotype, or physical appearance, and its genotype, or genetic makeup
- In the example of flower color in pea plants, PP and Pp plants have the same phenotype (purple) but different genotypes



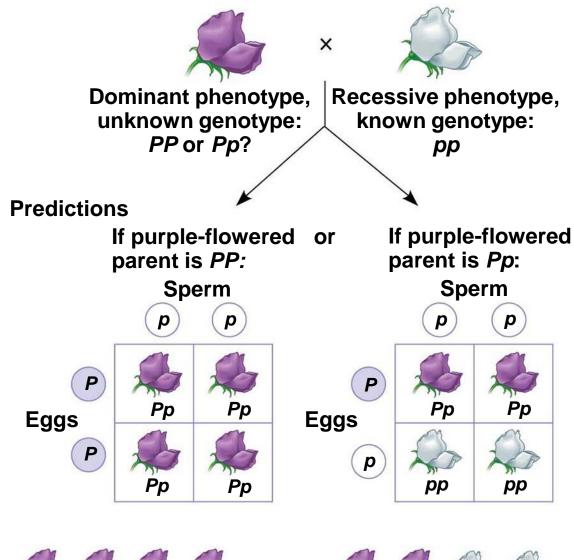
Ratio 3:1

Ratio 1:2:1

The Testcross

- How can we tell the genotype of an individual with the dominant phenotype?
- Such an individual could be either homozygous dominant or heterozygous
- The answer is to carry out a testcross: breeding the mystery individual with a homozygous recessive individual
- If any offspring display the recessive phenotype, the mystery parent must be heterozygous

Technique



Results



or



All offspring purple

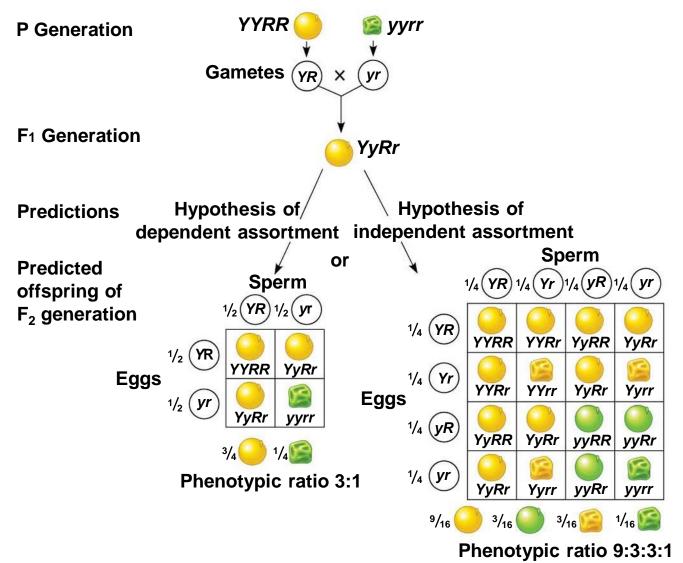
1/2 offspring purple and 1/2 offspring white

The Law of Independent Assortment

- Mendel derived the law of segregation by following a single character
- The F₁ offspring produced in this cross were monohybrids, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a monohybrid cross

- Mendel identified his second law of inheritance by following two characters at the same time
- Crossing two true-breeding parents differing in two characters produces dihybrids in the F₁ generation, heterozygous for both characters
- A dihybrid cross, a cross between F₁ dihybrids, can determine whether two characters are transmitted to offspring as a package or independently

Experiment

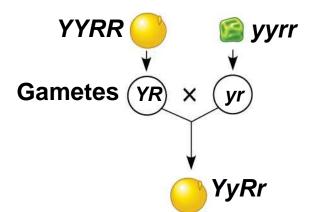


Results

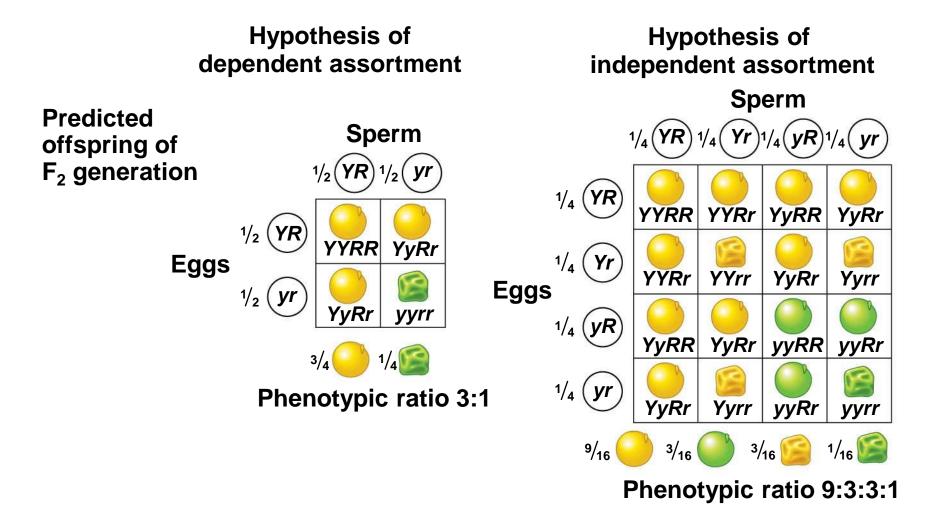
315 O 108 101 2 32 Phenotypic ratio approximately 9:3:3:1

Experiment

P Generation



F₁ Generation



Results

315



108



101



32

P

Phenotypic ratio approximately 9:3:3:1

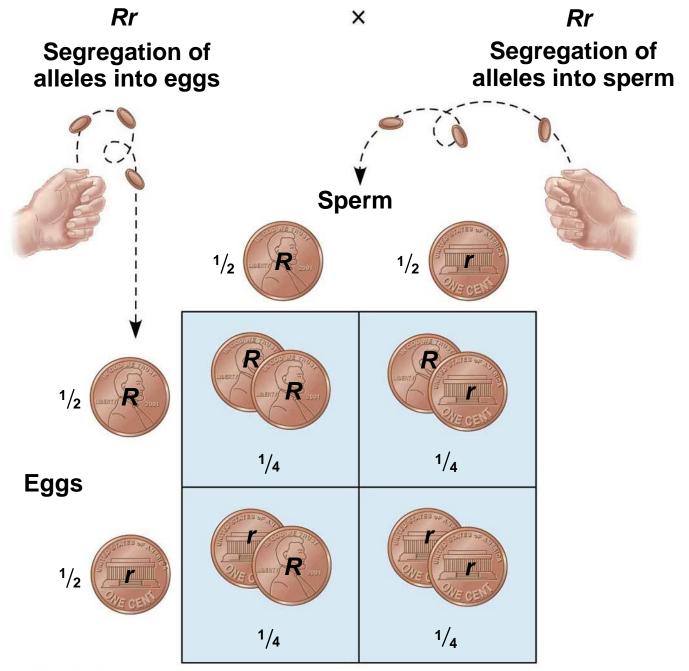
- The results of Mendel's dihybrid experiments are the basis for the law of independent assortment
- It states that each pair of alleles segregates independently of any other pair during gamete formation
- This law applies to genes on chromosomes that are not homologous, or those far apart on the same chromosome
- Genes located near each other on the same chromosome tend to be inherited together

Concept 11.2: Probability laws govern Mendelian inheritance

- Mendel's laws of segregation and independent assortment reflect the rules of probability
- The outcome of one coin toss has no impact on the outcome of the next toss
- In the same way, the alleles of one gene segregate into gametes independently of another gene's alleles

The Multiplication and Addition Rules Applied to Monohybrid Crosses

- The multiplication rule states that the probability that two or more independent events will occur together is the product of their individual probabilities
- This can be applied to an F₁ monohybrid cross
- Segregation in a heterozygous plant is like flipping a coin: Each gamete has a ½ chance of carrying the dominant allele and a ½ chance of carrying the recessive allele



- The addition rule states that the probability that any one of two or more mutually exclusive events will occur is calculated by adding together their individual probabilities
- It can be used to figure out the probability that an F₂ plant from a monohybrid cross will be heterozygous rather than homozygous

Solving Complex Genetics Problems with the Rules of Probability

- We can apply the rules of probability to predict the outcome of crosses involving multiple characters
- A dihybrid or other multicharacter cross is equivalent to two or more independent monohybrid crosses occurring simultaneously
- In calculating the chances for various genotypes, each character is considered separately, and then the individual probabilities are multiplied

 For example, if we cross F₁ heterozygotes of genotype YyRr, we can calculate the probability of different genotypes among the F₂ generation

Probability of YYRR =
$$\frac{1}{4}$$
 (probability of YY) $\times \frac{1}{4}$ (RR) = $\frac{1}{16}$

Probability of
$$YyRR = \frac{1}{2}(Yy) \times \frac{1}{4}(RR) = \frac{1}{8}$$

 For example, for the cross PpYyRr × Ppyyrr, we can calculate the probability of offspring showing at least two recessive traits

$$ppyyRr$$
 $\frac{1}{4}$ (probability of pp) × $\frac{1}{2}$ (yy) × $\frac{1}{2}$ (Rr) = $\frac{1}{16}$
 $ppYyrr$
 $\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$
 = $\frac{1}{16}$
 $Ppyyrr$
 $\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$
 = $\frac{2}{16}$
 $ppyyrr$
 $\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$
 = $\frac{1}{16}$
 $ppyyrr$
 $\frac{1}{4}$ × $\frac{1}{2}$ × $\frac{1}{2}$
 = $\frac{1}{16}$

Chance of at least two recessive traits = $\frac{6}{16}$ or $\frac{3}{8}$

Concept 11.3: Inheritance patterns are often more complex than predicted by simple Mendelian genetics

- Not all heritable characters are determined as simply as the traits Mendel studied
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance

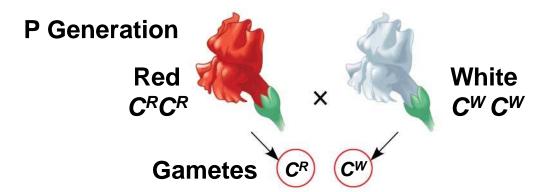
Extending Mendelian Genetics for a Single Gene

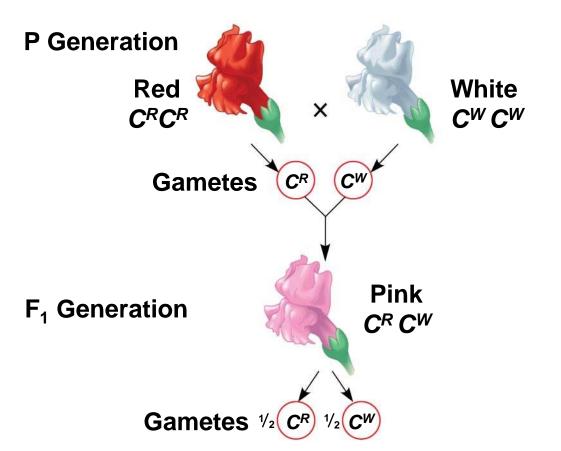
- Inheritance of characters by a single gene may deviate from simple Mendelian patterns in the following situations
 - When alleles are not completely dominant or recessive
 - When a gene has more than two alleles
 - When a single gene produces multiple phenotypes

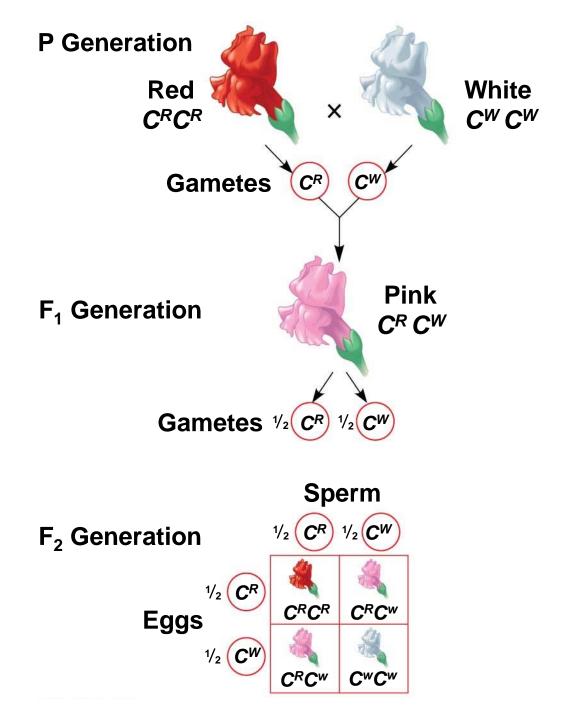
Degrees of Dominance

- Complete dominance occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In incomplete dominance, the phenotype of F₁
 hybrids is somewhere between the phenotypes of
 the two parental varieties
- In codominance, two dominant alleles affect the phenotype in separate, distinguishable ways

Figure 11.10-s1







The Relationship Between Dominance and Phenotype

- Alleles are simply variations in a gene's nucleotide sequence
- When a dominant allele coexists with a recessive allele in a heterozygote, they do not actually interact at all
- For any character, dominant/recessive relationships of alleles depend on the level at which we examine the phenotype

- Tay-Sachs disease is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
 - At the organismal level, the allele is recessive
 - At the biochemical level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
 - At the molecular level, the alleles are codominant

Frequency of Dominant Alleles

- Dominant alleles are not necessarily more common in populations than recessive alleles
- For example, one baby out of 400 in the United States is born with extra fingers or toes, a dominant trait called polydactyly

Multiple Alleles

- Most genes exist in populations in more than two allelic forms
- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles of the gene: I^A, I^B, and i.
- The enzyme (I) adds specific carbohydrates to the surface of blood cells
- The enzyme encoded by I^A adds the A carbohydrate, and the enzyme encoded by I^B adds the B carbohydrate; the enzyme encoded by the i allele adds neither

(a) The three alleles for the ABO blood groups and their carbohydrates **Allele J**B ĮΑ Carbohydrate $\mathsf{B} \bigcirc$ $A \triangle$ none (b) Blood group genotypes and phenotypes $I^A I^A$ or $I^A i \mid I^B I^B$ or $I^B i$ Genotype ii **J**A **J**B Red blood cell appearance **Phenotype** В AB (blood group)

Pleiotropy

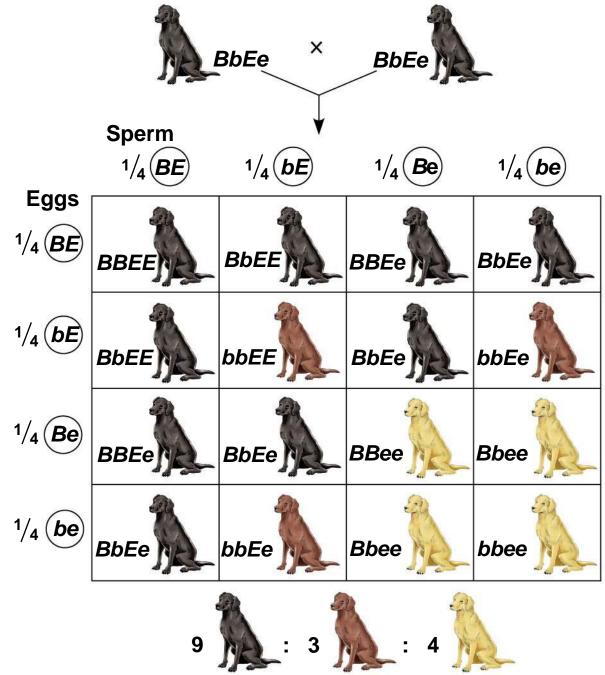
- Most genes have multiple phenotypic effects, a property called pleiotropy
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease

Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes
- The gene products may interact
- Alternatively, multiple genes could independently affect a single trait

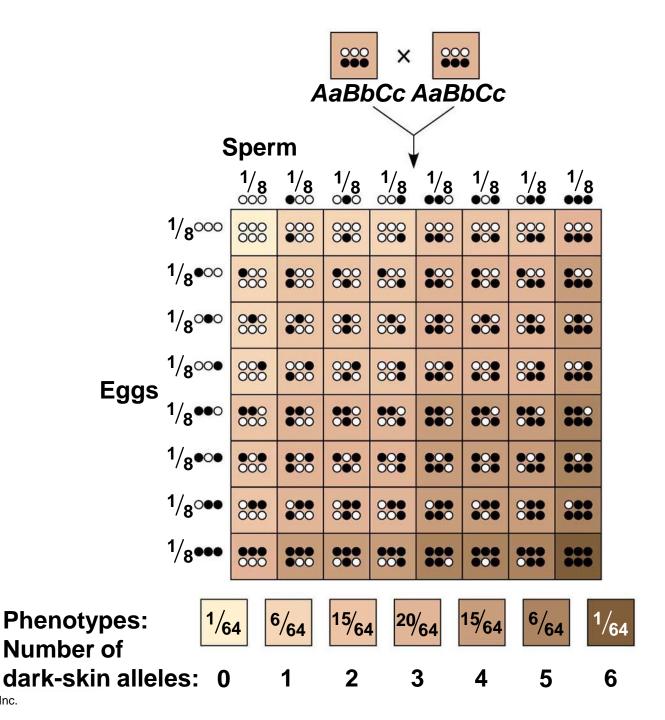
Epistasis

- In epistasis, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes
- One gene determines the pigment color (with alleles B for black and b for brown)
- The other gene (with alleles C for color and c for no color) determines whether the pigment will be deposited in the hair



Polygenic Inheritance

- Quantitative characters are those that vary in the population along a continuum
- Quantitative variation usually indicates polygenic inheritance, an additive effect of two or more genes on a single phenotype
- Skin color in humans is an example of polygenic inheritance



© 2016 Pearson Education, Inc.

Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype
- The norm of reaction is the phenotypic range of a genotype influenced by the environment

- The phenotypic range is generally broadest for polygenic characters
- Such characters are called multifactorial because genetic and environmental factors collectively influence phenotype

A Mendelian View of Heredity and Variation

- An organism's phenotype includes its physical appearance, internal anatomy, physiology, and behavior
- An organism's phenotype reflects its overall genotype and unique environmental history

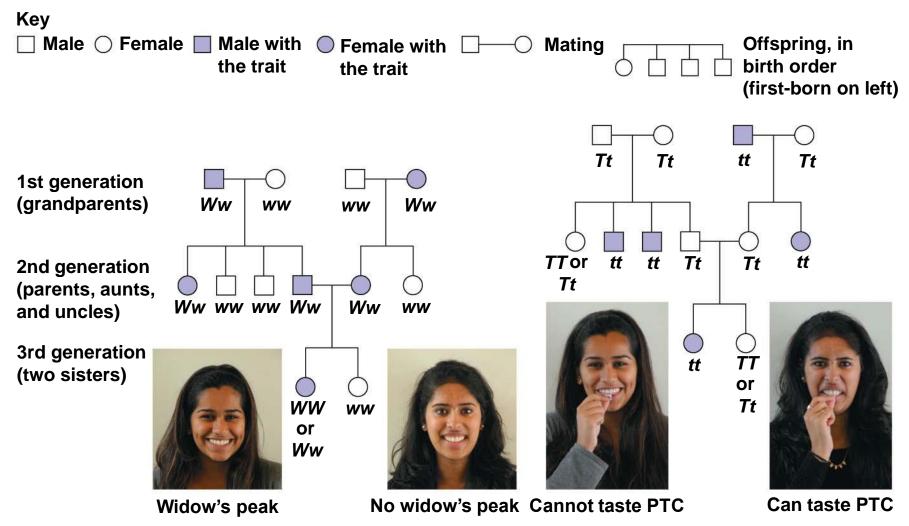
Concept 11.4: Many human traits follow Mendelian patterns of inheritance

- Humans are not good subjects for genetic research
 - Generation time is too long
 - Parents produce relatively few offspring
 - Breeding experiments are unacceptable
- However, basic Mendelian genetics endures as the foundation of human genetics

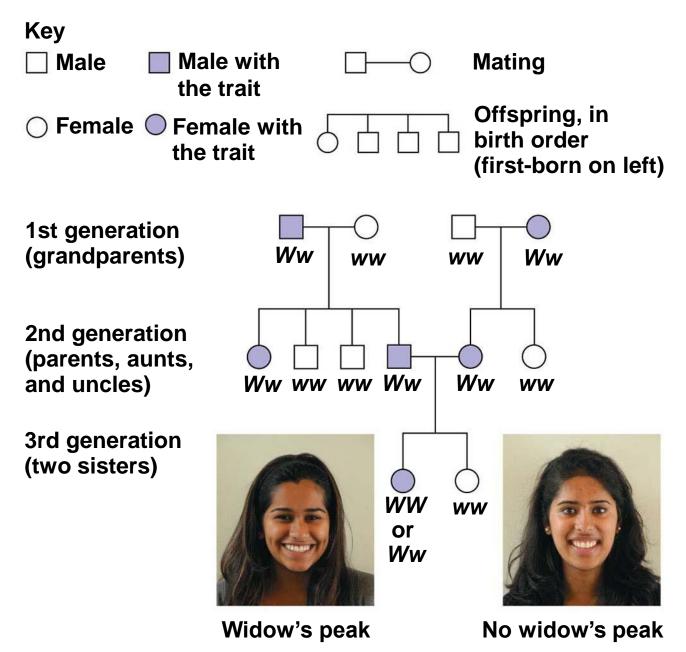
Pedigree Analysis

- A pedigree is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees

- Pedigrees can also be used to make predictions about future offspring
- We can use the multiplication and addition rules to predict the probability of specific phenotypes



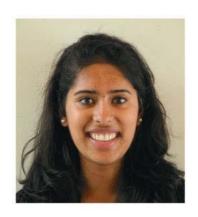
- (a) Is a widow's peak a dominant or recessive trait?
- (b) Is the inability to taste a chemical called PTC a dominant or recessive trait?



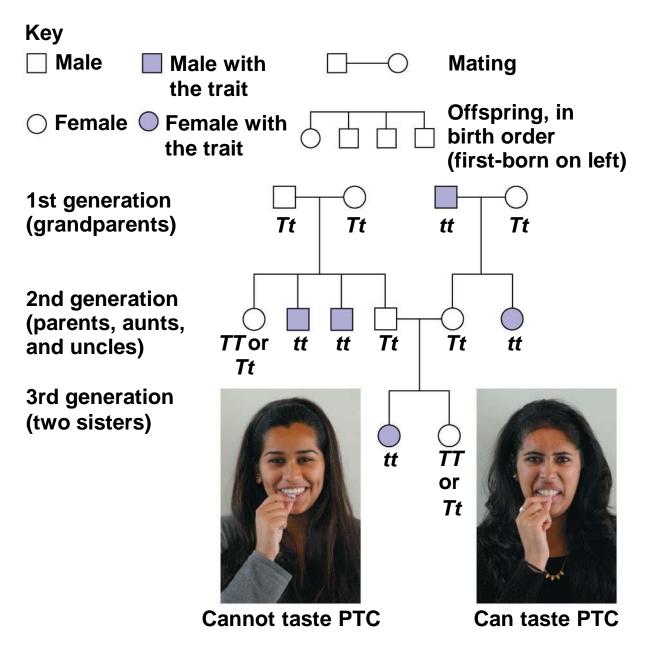
(a) Is a widow's peak a dominant or recessive trait?



Widow's peak



No widow's peak



(b) Is the inability to taste a chemical called PTC a dominant or recessive trait?



Cannot taste PTC



Can taste PTC

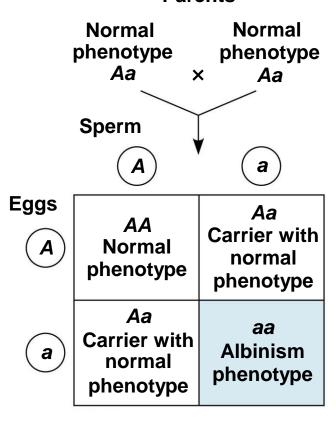
Recessively Inherited Disorders

- Many genetic disorders are inherited in a recessive manner
- These range from relatively mild to life-threatening

The Behavior of Recessive Alleles

- Recessively inherited disorders show up only in individuals homozygous for the allele
- Carriers are heterozygous individuals who carry the recessive allele but are phenotypically normal
- Most people who have recessive disorders are born to parents who are carriers of the disorder

Parents







- If a recessive allele that causes a disease is rare, then the chance of two carriers meeting and mating is low
- Consanguineous (between close relatives) matings increase the chance of mating between two carriers of the same rare allele
- Most societies and cultures have laws or taboos against marriages between close relatives

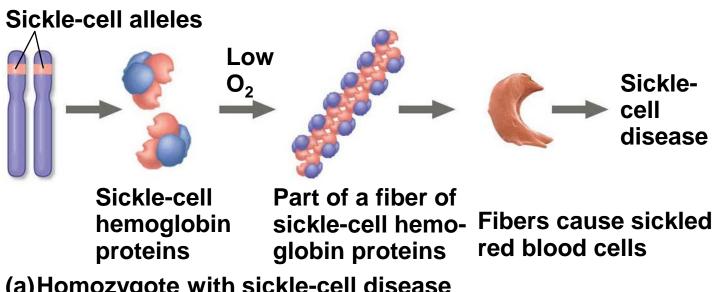
Cystic Fibrosis

- Cystic fibrosis is the most common lethal genetic disease in the United States, striking one out of every 2,500 people of European descent
- The cystic fibrosis allele results in defective or absent chloride transport channels in plasma membranes leading to a buildup of chloride ions outside the cell
- Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine

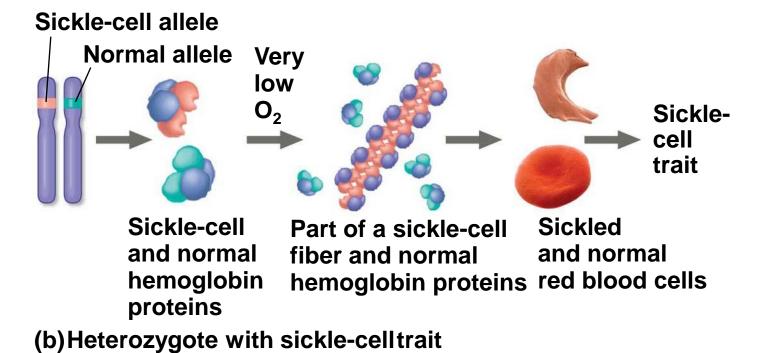
Sickle-Cell Disease: A Genetic Disorder with Evolutionary Implications

- Sickle-cell disease affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- In homozygous individuals, all hemoglobin is abnormal (sickle-cell)
- Symptoms include physical weakness, pain, organ damage, and even stroke and paralysis

- Heterozygotes (said to have sickle-cell trait) are usually healthy but may suffer some symptoms
- About one out of ten African-Americans has sicklecell trait, an unusually high frequency of an allele with detrimental effects in homozygotes
- Heterozygotes are less susceptible to the malaria parasite, so there is an advantage to being heterozygous



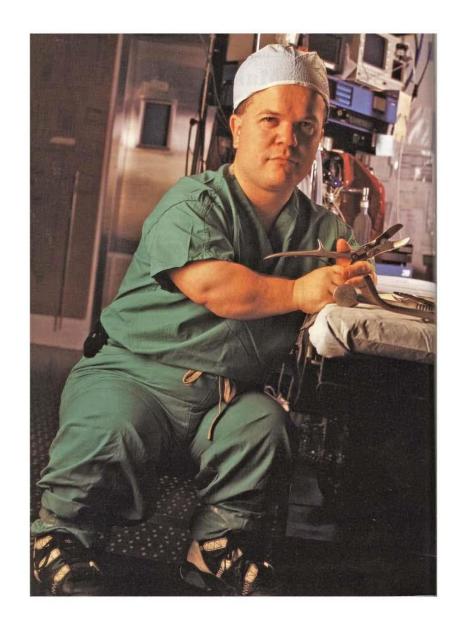
(a) Homozygote with sickle-cell disease

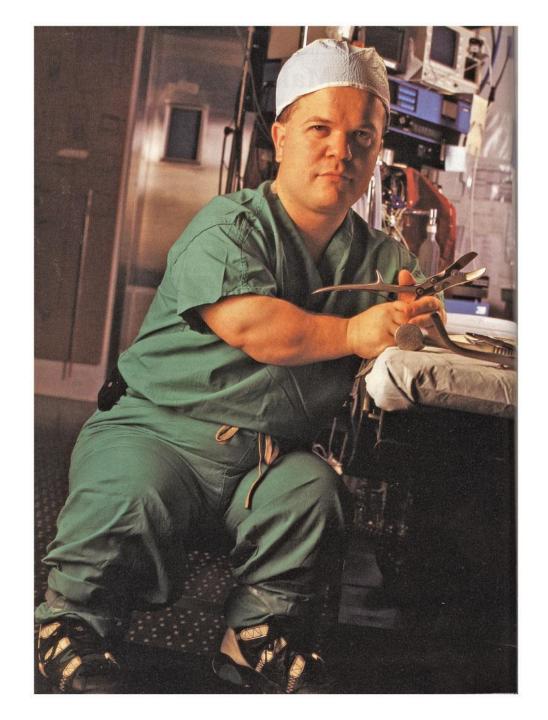


Dominantly Inherited Disorders

- Some human disorders are caused by dominant alleles
- Dominant alleles that cause a lethal disease are rare and arise by mutation
- Achondroplasia is a form of dwarfism caused by a rare dominant allele

Parents Normal Dwarf × phenotype phenotype Dd dd **Sperm Eggs** Dd dd **Normal Dwarf** phenotype phenotype Dd dd **Normal Dwarf** d phenotype phenotype





- The timing of onset of a disease significantly affects its inheritance
- Huntington's disease is a degenerative disease of the nervous system
- The disease has no obvious phenotypic effects until the individual is about 35 to 45 years of age
- Once the deterioration of the nervous system begins the condition is irreversible and fatal

Multifactorial Disorders

- Many diseases, such as heart disease, diabetes, alcoholism, mental illnesses, and cancer, have both genetic and environmental components
- Lifestyle has a tremendous effect on phenotype for cardiovascular health and other multifactorial characters

Genetic Counseling Based on Mendelian Genetics

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease
- Each child represents an independent event in the sense that its genotype is unaffected by the genotypes of older siblings

Phenotypes:

1/64

6/64

15/64

20/64

15/64

Number of

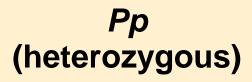
dark-skin alleles: 0

6





PP (homozygous)





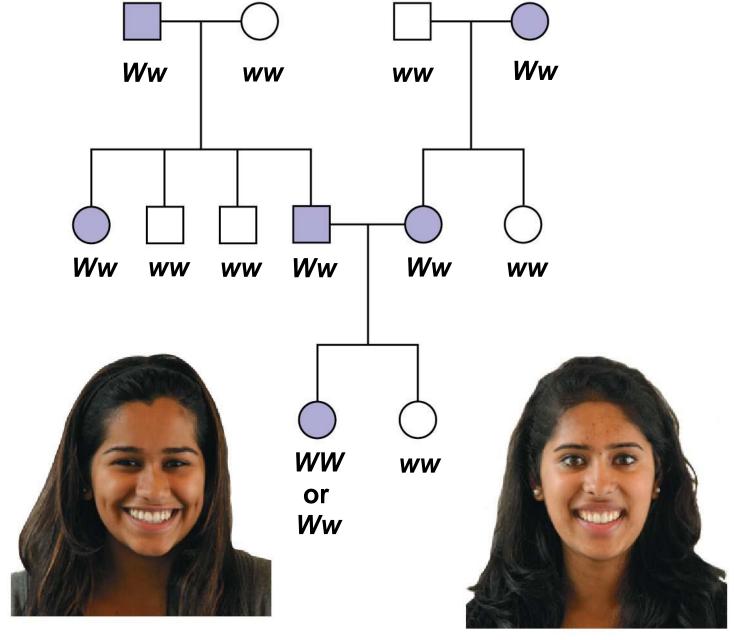
Pp (heterozygous)



pp (homozygous)

Relationship among alleles of a single gene	Description	Example
Complete dominance of one allele	Heterozygous phenotype same as that of homo-zygous dominant	PP Pp
Incomplete dominance of either allele	Heterozygous phenotype intermediate between the two homozygous phenotypes	CR CR CR CW CW CW
Codominance	Both phenotypes expressed in heterozygotes	IA IB
Multiple alleles	In the population, some genes have more than two alleles	ABO blood group alleles
Pleiotropy	One gene affects multiple phenotypic characters	Sickle-cell disease

Relationship among two or more genes	Description	Example
Epistasis	The phenotypic expression of one gene affects the expression of another gene	BbEe X BbEe BE BE BE Be be Be Be Be Be Be Be Be Be Be Be Be Be Be B
Polygenic inheritance	A single phenotypic character is affected by two or more genes	AaBbCc



Widow's peak

No widow's peak

