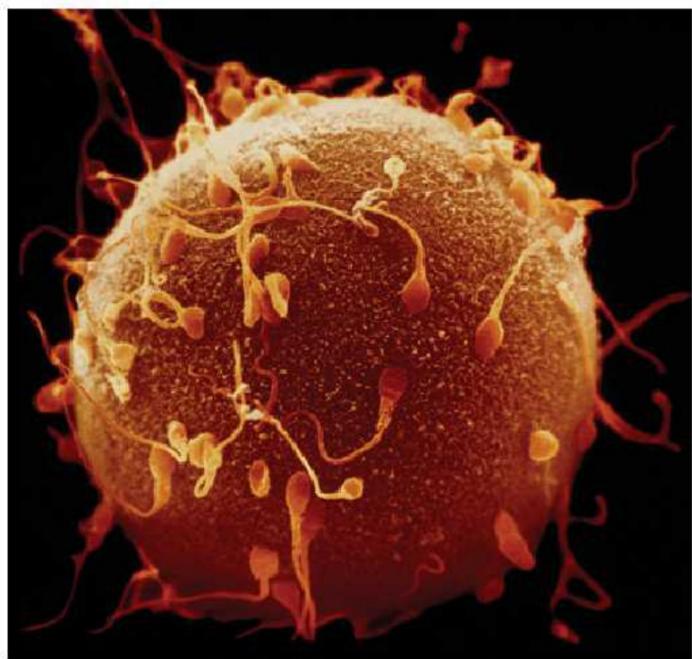




## 6.3 Mendel and Heredity

### KEY CONCEPT

Mendel's research showed that traits are inherited as discrete units.



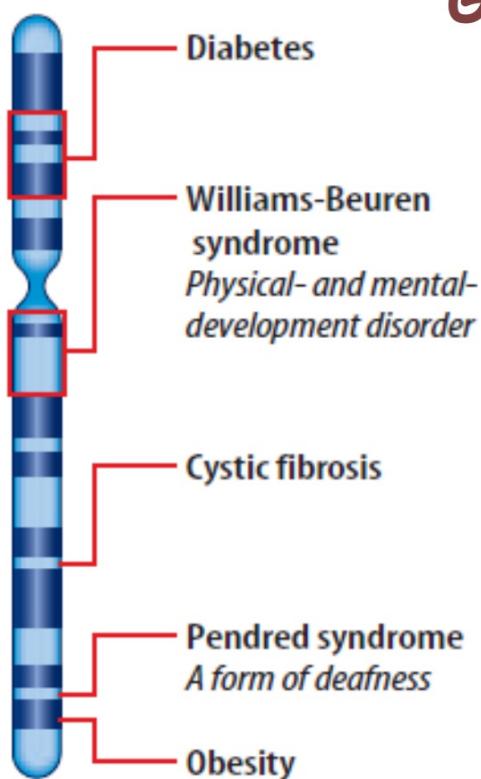
## 6.3 Mendel and Heredity

### • Mendel laid the groundwork for genetics.

- Traits are distinguishing characteristics that are inherited.
- Genetics is the study of biological inheritance patterns and variation.
- Gregor Mendel showed that traits are inherited as discrete units.
- Many in Mendel's day thought traits were blended.



## Chromosome 7



## Genes on chromosomes

**Table 2 Principles of Heredity**

1	Traits are controlled by alleles on chromosomes.
2	An allele's effect is dominant or recessive.
3	When a pair of chromosomes separates during meiosis, the different alleles for a trait move into separate sex cells.



**Alleles are alternate forms of the same gene. They can be dominant or recessive.**

[http://www.ornl.gov/sci/techresources/Human\\_Genome/posters/chromosome/chooser.shtml](http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml)

# X

Short stature; idiopathic familial  
 Leri-Weill dyschondrosteosis  
 Langer mesomelic dysplasia  
 Leukemia, acute myeloid, M2 type  
 Chondrodysplasia punctata  
 Kallmann syndrome  
 Ocular albinism, Nettleship-Falls type  
 Oral-facial-digital syndrome  
 Nance-Horan cataract-dental syndrome  
 Heterocellular hereditary persistence of fetal hemoglobin  
 Pyruvate dehydrogenase deficiency  
 Glycogen storage disease  
 Coffin-Lowry syndrome  
 Mental retardation  
 Spondyloepiphyseal dysplasia tarda  
 Paroxysmal nocturnal hemoglobinuria  
 Infantile spasmodic syndrome  
 Aicardi syndrome  
 Deafness, sensorineural  
 Simpson-Golabi-Behmel syndrome, type 2  
 Adrenal hypoplasia, congenital  
 Dosage-sensitive sex reversal  
 Deafness, congenital sensorineural  
 Retinitis pigmentosa  
 Wilson-Turner syndrome  
 Cone dystrophy  
 Aland island eye disease (ocular albinism)  
 Optic atrophy  
 Night blindness, congenital stationary, type 1  
 Erythroid-potentiating activity  
 Arthrogryposis multiplex congenita  
 Night blindness, congenital stationary, type 2  
 Brunner syndrome  
 Wiskott-Aldrich syndrome  
 Thrombocytopenia  
 Dent disease  
 Nephrolithiasis, type I  
 Hypophosphatasuria, type III  
 Proteinuria  
 Anemia, sideroblastic/hypochromic  
 Cerebellar ataxia  
 Renal cell carcinoma, papillary  
 Diabetes mellitus, insulin-dependent  
 Sutherland-Haan syndrome  
 Cognitive function, social

## 153 million base pairs



Hodgkin disease susceptibility, pseudoautosomal  
 Ichthyosis  
 Microphthalmia, dermal aplasia, and sclerocornea  
 Episodic muscle weakness  
 Mental retardation  
 Ocular albinism and sensorineural deafness  
 Amelogenesis imperfecta  
 Charcot-Marie-Tooth disease, recessive  
 Keratosis follicularis spinulosa decalvans  
 Hypophosphatasia, hereditary  
 Partington syndrome  
 Retinoschisis  
 Gonadal dysgenesis, XY female type  
 Mental retardation, non-dysmorphic  
 Agammaglobulinemia, type 2  
 Craniofrontonasal dysplasia  
 Opitz G syndrome, type I  
 Pigment disorder, reticulate  
 Melanoma  
 Duchenne muscular dystrophy  
 Becker muscular dystrophy  
 Cardiomyopathy, dilated  
 Chronic granulomatous disease  
 Snyder-Robinson mental retardation  
 Norrie disease  
 Exudative vitreoretinopathy  
 Coats disease  
 Renshaw syndrome  
 Retinitis pigmentosa, recessive  
 Mental retardation, nonspecific and syndromic  
 Dyserythropoietic anemia with thrombocytopenia  
 Chondrodysplasia punctata, dominant  
 Autoimmunity-immunodeficiency syndrome  
 Renal cell carcinoma, papillary  
 Facio-genital dysplasia (Aarskog-Scott syndrome)  
 Chorioathetosis with mental retardation  
 Sarcoma, synovial  
 Prieto syndrome  
 Spinal muscular atrophy, lethal infantile  
 Migraine, familial typical  
 Androgen insensitivity  
 Spinal and bulbar muscular atrophy  
 Prostate cancer  
 Perineal hypospadias  
 Breast cancer, male, with Reifenstein syndrome

# Y

## 50 million base pairs

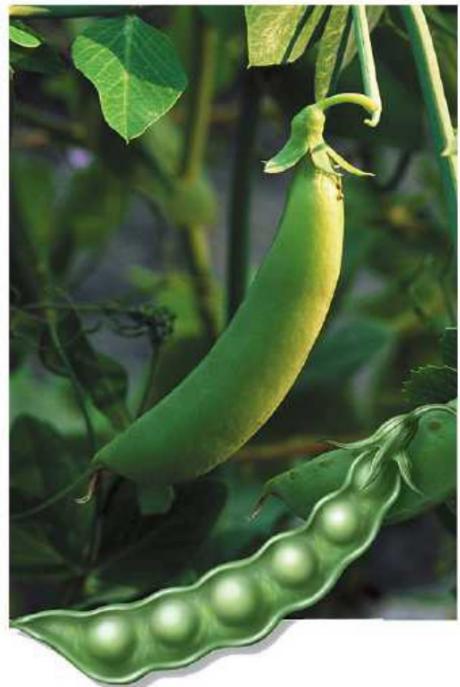


Short stature homeobox, Y-linked  
 Short stature  
 Leri-weill dyschondrosteosis  
 Langer mesomelic dysplasia  
 Interleukin-3 receptor, Y chromosomal  
 Sex-determining region Y (testis-determining)  
 Gonadal dysgenesis, XY type  
 Protocadherin 11, Y-linked  
 Azoospermia factors  
 Male infertility due to spermatogenic failure  
 Growth control, Y-chromosome influenced  
 Chromodomain proteins  
 Retinitis pigmentosa, Y-linked

## 6.3 Mendel and Heredity

### • Mendel's data revealed patterns of inheritance.

- Mendel made three key decisions in his experiments.
  - use of purebred plants
  - control over breeding
  - observation of seven “either-or” traits



## 6.3 Mendel and Heredity

- Mendel used pollen to fertilize selected pea plants.
  - P generation crossed to produce F<sub>1</sub> generation
  - interrupted the self-pollination process by removing male flower parts



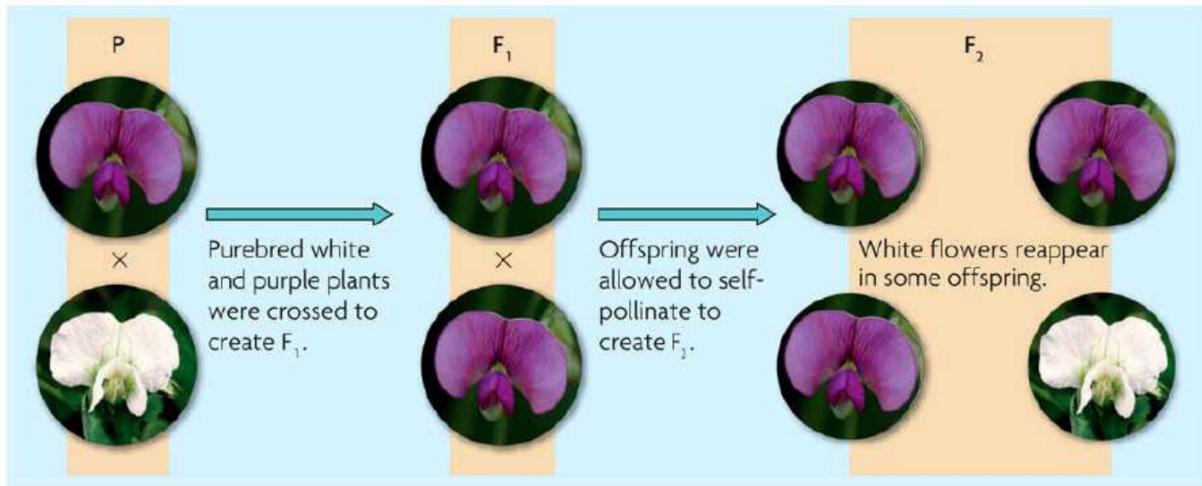
Mendel controlled the fertilization of his pea plants by removing the male parts, or stamens.



He then fertilized the female part, or pistil, with pollen from a different pea plant.

## 6.3 Mendel and Heredity

- Mendel allowed the resulting plants to self-pollinate.
  - Among the F<sub>1</sub> generation, all plants had purple flowers
  - F<sub>1</sub> plants are all heterozygous
  - Among the F<sub>2</sub> generation, some plants had purple flowers and some had white



## 6.3 Mendel and Heredity

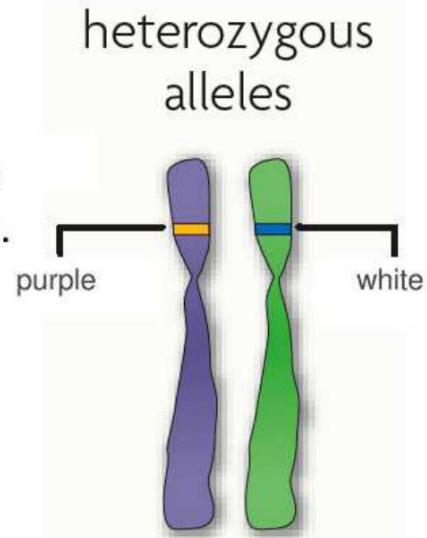
- Mendel observed patterns in the first and second generations of his crosses.

**FIGURE 6.10 MENDEL'S MONOHYBRID CROSS RESULTS**

F <sub>2</sub> TRAITS	DOMINANT	RECESSIVE	RATIO
Pea shape	5474 round	1850 wrinkled	2.96:1
Pea color	6022 yellow	2001 green	3.01:1
Flower color	705 purple	224 white	3.15:1
Pod shape	882 smooth	299 constricted	2.95:1
Pod color	428 green	152 yellow	2.82:1
Flower position	651 axial	207 terminal	3.14:1
Plant height	787 tall	277 short	2.84:1

## 6.3 Mendel and Heredity

- Mendel drew three important conclusions.
  - Traits are inherited as discrete units.
  - Organisms inherit two copies of each gene, one from each parent.
  - The two copies segregate during gamete formation.
  - The last two conclusions are called the law of segregation.



## 6.4 Traits, Genes, and Alleles

### KEY CONCEPT

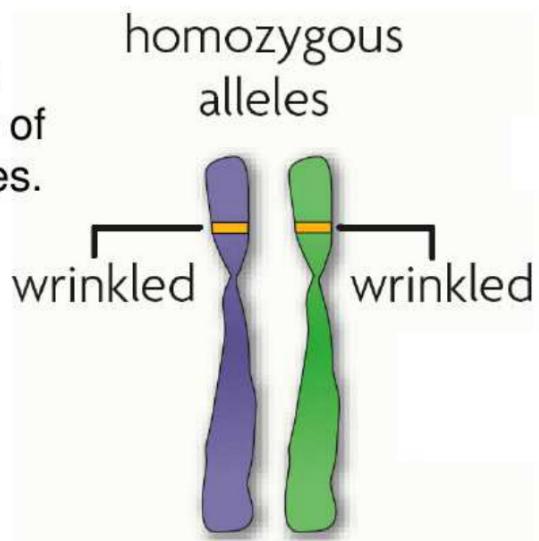
Genes encode proteins that produce a diverse range of traits.



## 6.4 Traits, Genes, and Alleles

### • The same gene can have many versions.

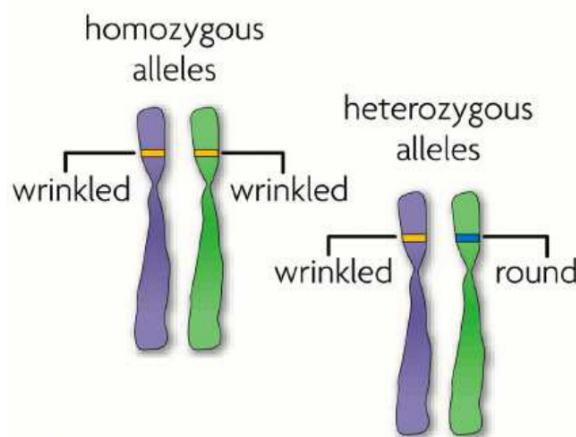
- A gene is a piece of DNA that directs a cell to make a certain protein.
- Each gene has a locus, a specific position on a pair of homologous chromosomes.



## 6.4 Traits, Genes, and Alleles

- An allele is any alternative form of a gene occurring at a specific locus on a chromosome.
  - Each parent donates one allele for every gene.
  - Homozygous describes two alleles that are the same at a specific locus.
  - Heterozygous describes two alleles that are different at a specific locus.

**Homozygous** alleles are identical to each other.



**Heterozygous** alleles are different from each other.

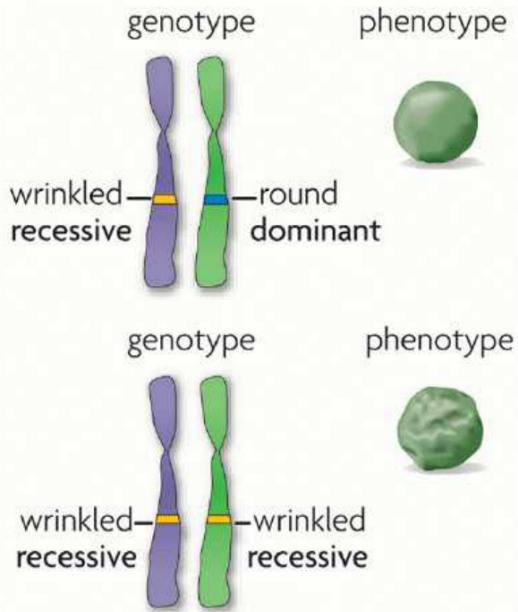
## 6.4 Traits, Genes, and Alleles

### ➲ Genes influence the development of traits.

- All of an organism's genetic material is called the genome.
- A genotype refers to the makeup of a specific set of genes.
- A phenotype is the physical expression of a trait.

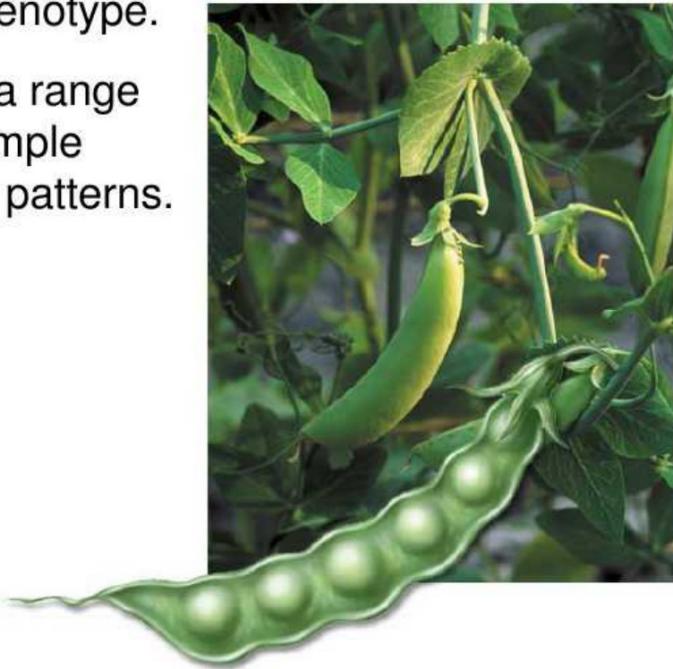
## 6.4 Traits, Genes, and Alleles

- Alleles can be represented using letters.
  - A dominant allele is expressed as a phenotype when at least one allele is dominant.
  - A recessive allele is expressed as a phenotype only when two copies are present.
  - Dominant alleles are represented by uppercase letters; recessive alleles by lowercase letters.



## 6.4 Traits, Genes, and Alleles

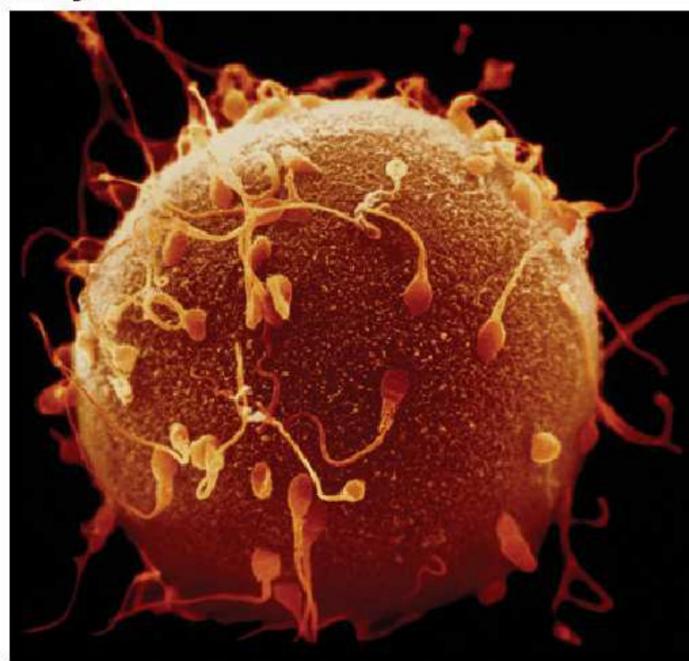
- Both homozygous dominant and heterozygous genotypes yield a dominant phenotype.
- Most traits occur in a range and do not follow simple dominant-recessive patterns.



## 6.5 Traits and Probability

### KEY CONCEPT

The inheritance of traits follows the rules of probability.

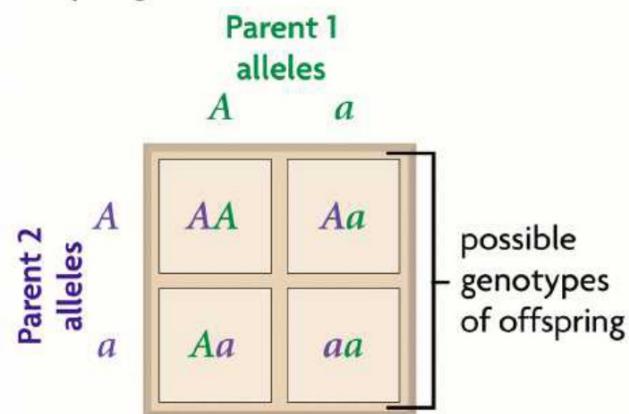


## 6.5 Traits and Probability

### • Punnett squares illustrate genetic crosses.

- The Punnett square is a grid system for predicting all possible genotypes resulting from a cross.
  - The axes represent the possible gametes of each parent.
  - The boxes show the possible genotypes of the offspring.
- The Punnett square yields the ratio of possible genotypes and phenotypes.

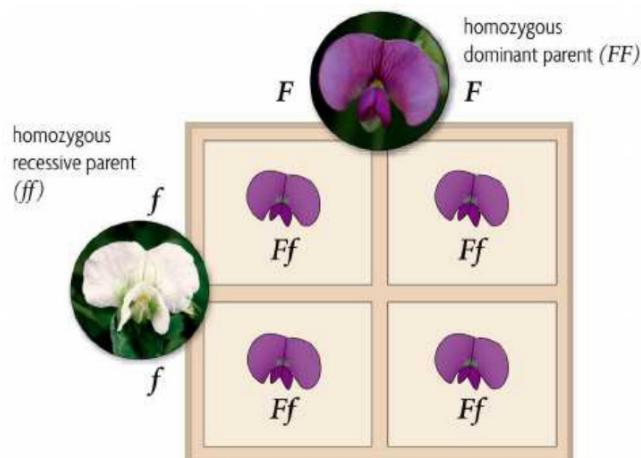
The **Punnett square** is a grid system for predicting possible genotypes of offspring.



## 6.5 Traits and Probability

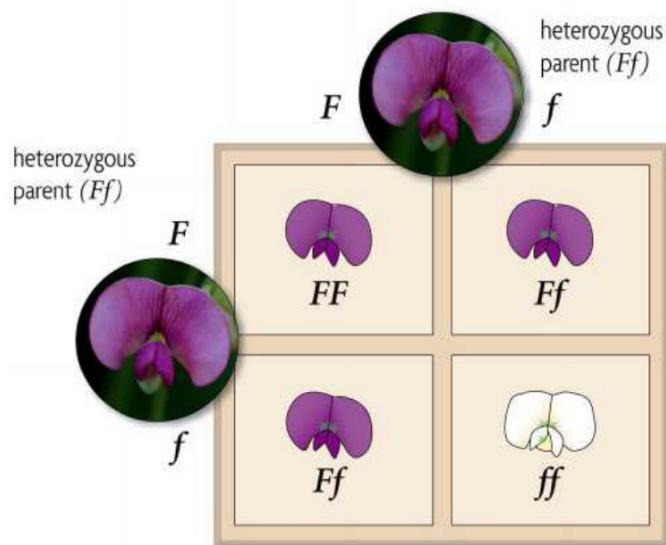
### • A monohybrid cross involves one trait.

- Monohybrid crosses examine the inheritance of only one specific trait.
  - homozygous dominant-homozygous recessive: all heterozygous, all dominant



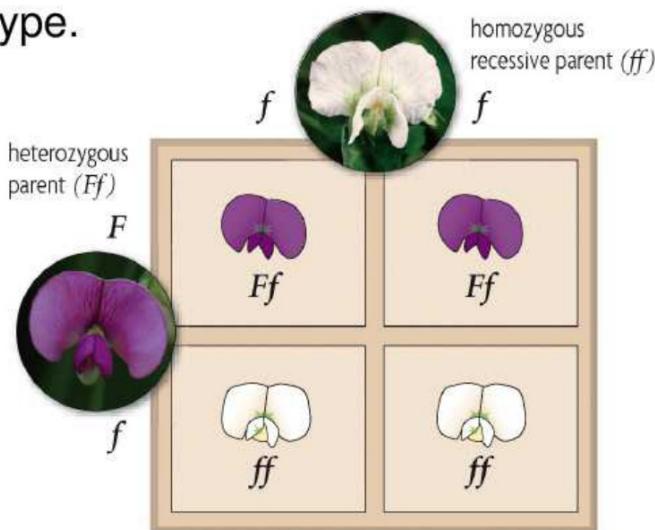
## 6.5 Traits and Probability

- heterozygous-heterozygous—1:2:1 homozygous dominant: heterozygous:homozygous recessive; 3:1 dominant:recessive



## 6.5 Traits and Probability

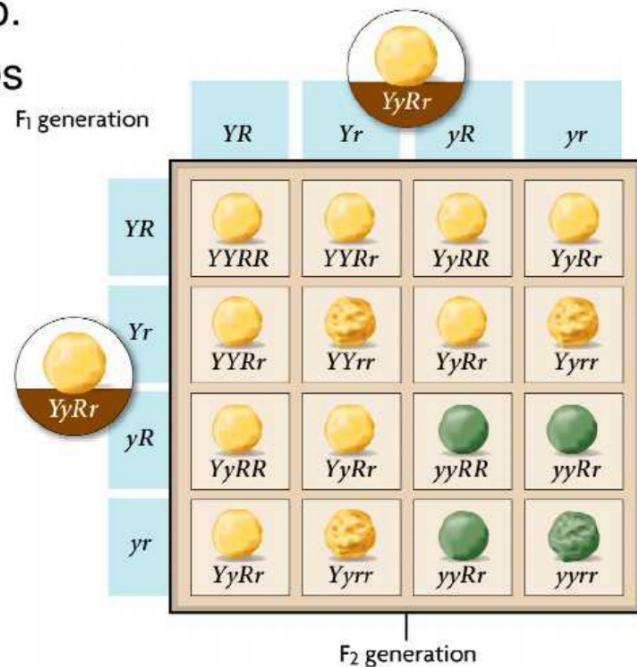
- heterozygous-homozygous recessive—1:1  
heterozygous:homozygous recessive; 1:1  
dominant:recessive
- A testcross is a cross between an organism with an unknown genotype and an organism with the recessive phenotype.



## 6.5 Traits and Probability

### ► A dihybrid cross involves two traits.

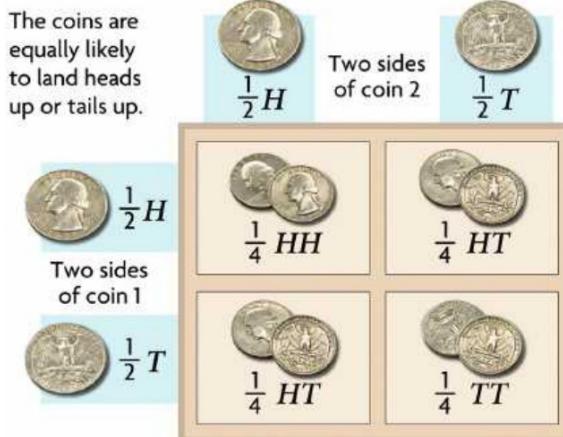
- Mendel's dihybrid crosses with heterozygous plants yielded a 9:3:3:1 phenotypic ratio.
- Mendel's dihybrid crosses led to his second law, the law of independent assortment.
- The law of independent assortment states that allele pairs separate independently of each other during meiosis.



## 6.5 Traits and Probability

### • Heredity patterns can be calculated with probability.

- Probability is the likelihood that something will happen.
- Probability predicts an average number of occurrences, not an exact number of occurrences.
- Probability =  $\frac{\text{number of ways a specific event can occur}}{\text{number of total possible outcomes}}$
- Probability applies to random events such as meiosis and fertilization.



## 7.1 Chromosomes and Phenotype

### KEY CONCEPT

The chromosomes on which genes are located can affect the expression of traits.



## 7.1 Chromosomes and Phenotype

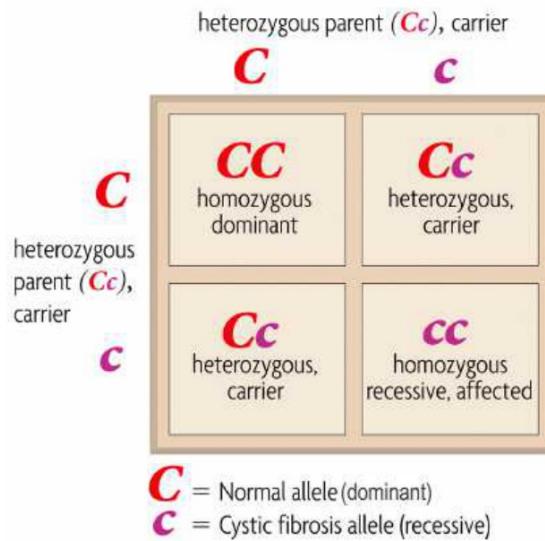
### • Two copies of each autosomal gene affect phenotype.

- Mendel studied autosomal gene traits, like hair texture.



## 7.1 Chromosomes and Phenotype

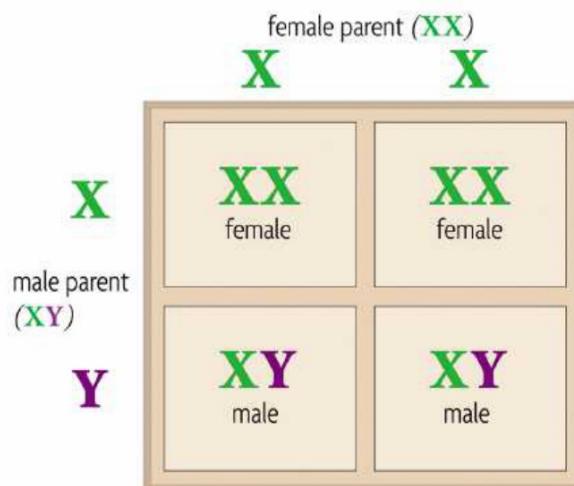
- Mendel's rules of inheritance apply to autosomal genetic disorders.
  - A heterozygote for a recessive disorder is a carrier.
  - Disorders caused by dominant alleles are uncommon.



## 7.1 Chromosomes and Phenotype

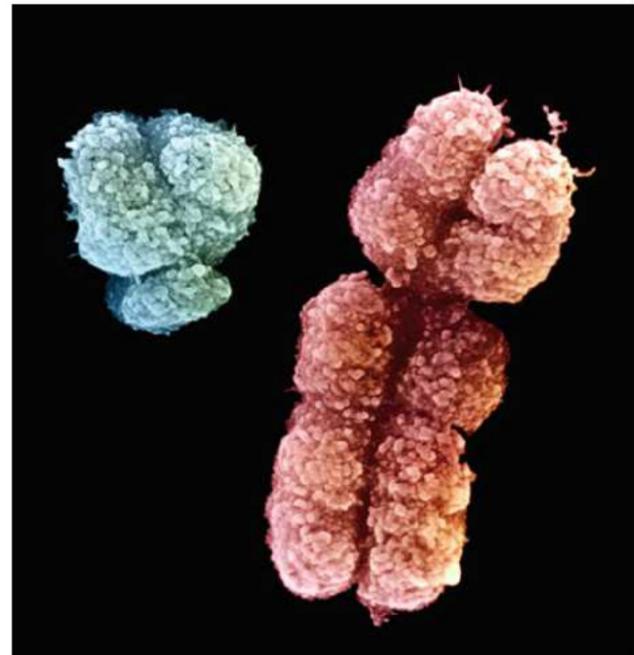
### • Males and females can differ in sex-linked traits.

- Genes on sex chromosomes are called sex-linked genes.
  - Y chromosome genes in mammals are responsible for male characteristics.
  - X chromosome genes in mammals affect many traits.



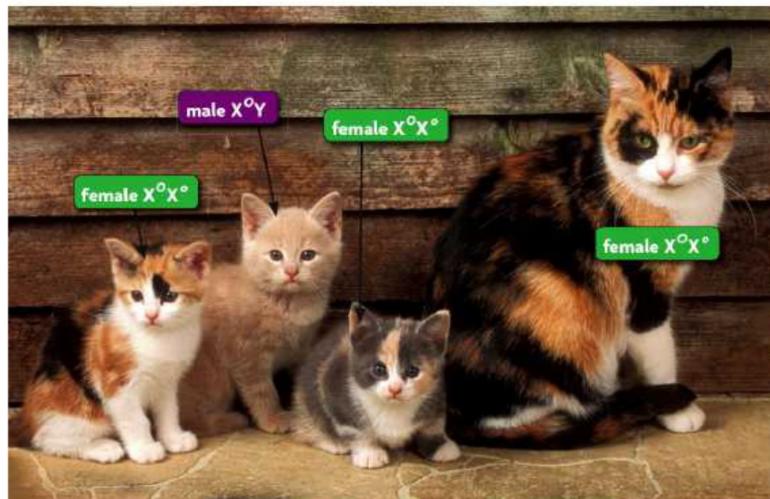
## 7.1 Chromosomes and Phenotype

- Male mammals have an XY genotype.
  - All of a male's sex-linked genes are expressed.
  - Males have no second copies of sex-linked genes.



## 7.1 Chromosomes and Phenotype

- Female mammals have an XX genotype.
  - Expression of sex-linked genes is similar to autosomal genes in females.
  - X chromosome inactivation randomly “turns off” one X chromosome.



## 7.2 Complex Patterns of Inheritance

### KEY CONCEPT

**Phenotype is affected by many different factors.**



## 7.2 Complex Patterns of Inheritance

- **Phenotype can depend on interactions of alleles.**

- In incomplete dominance, neither allele is completely dominant nor completely recessive.
  - Heterozygous phenotype is intermediate between the two homozygous phenotypes
  - Homozygous parental phenotypes not seen in  $F_1$  offspring



## 7.2 Complex Patterns of Inheritance

- Codominant alleles will both be completely expressed.
  - Codominant alleles are neither dominant nor recessive.
  - The ABO blood types result from codominant alleles.
- Many genes have more than two alleles.

PHENOTYPE (BLOOD TYPE)		GENOTYPES
A	antigen A 	$I^A I^A$ or $I^A i$
B		$I^B I^B$ or $I^B i$
AB	both antigens 	$I^A I^B$
O	no antigens 	$ii$

## 7.2 Complex Patterns of Inheritance

- Many genes may interact to produce one trait.

- Polygenic traits are produced by two or more genes.



Order of dominance:  
brown > green > blue.

GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue

## 7.2 Complex Patterns of Inheritance

- An epistatic gene can interfere with other genes.



## 7.2 Complex Patterns of Inheritance

### • The environment interacts with genotype.

- Phenotype is a combination of genotype and environment.
- The sex of sea turtles depends on both genes and the environment
- Height is an example of a phenotype strongly affected by the environment.



## 7.4 Human Genetics and Pedigrees

### KEY CONCEPT

A combination of methods is used to study human genetics.



## 7.4 Human Genetics and Pedigrees

- **Human genetics follows the patterns seen in other organisms.**

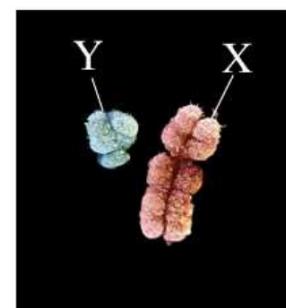
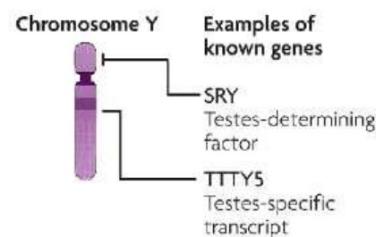
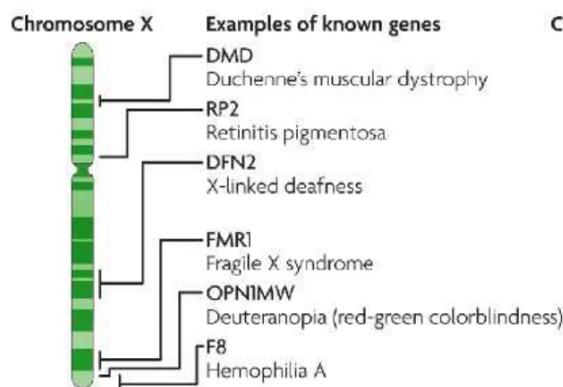
- The basic principles of genetics are the same in all sexually reproducing organisms.
  - Inheritance of many human traits is complex.
  - Single-gene traits are important in understanding human genetics.



## 7.4 Human Genetics and Pedigrees

### • Females can carry sex-linked genetic disorders.

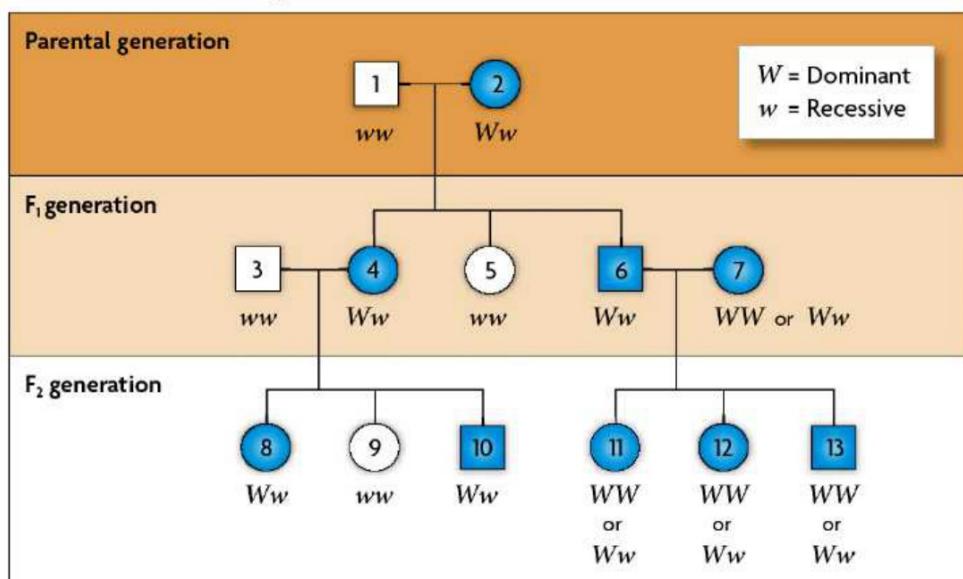
- Males (XY) express all of their sex linked genes.
- Expression of the disorder depends on which parent carries the allele and the sex of the child.



## 7.4 Human Genetics and Pedigrees

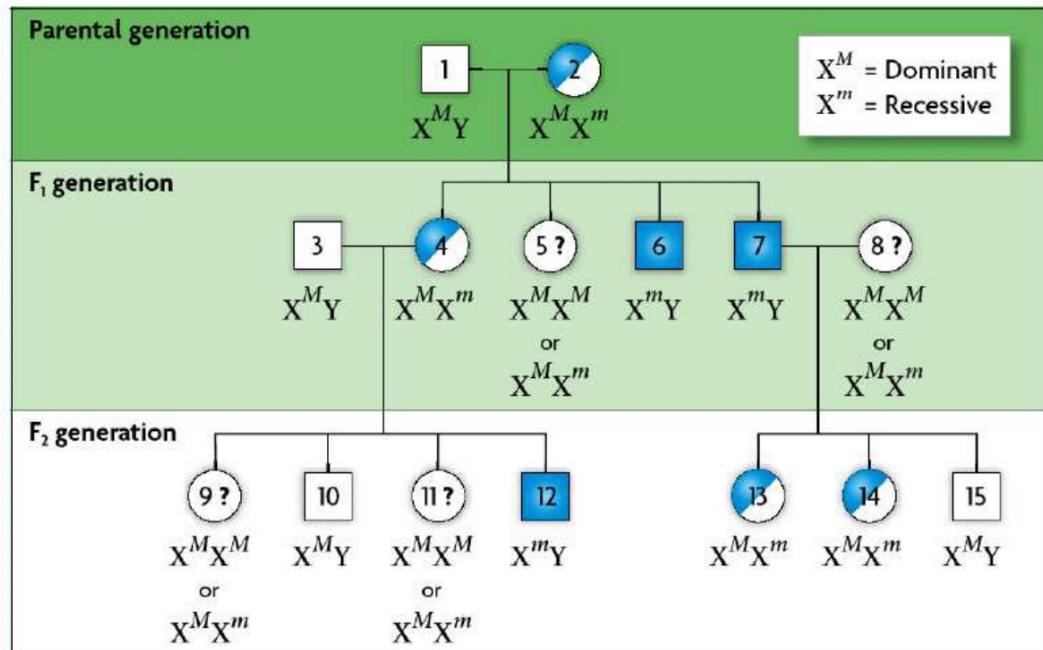
- A pedigree is a chart for tracing genes in a family.

- Phenotypes are used to infer genotypes on a pedigree.
- Autosomal genes show different patterns on a pedigree than sex-linked genes.



## 7.4 Human Genetics and Pedigrees

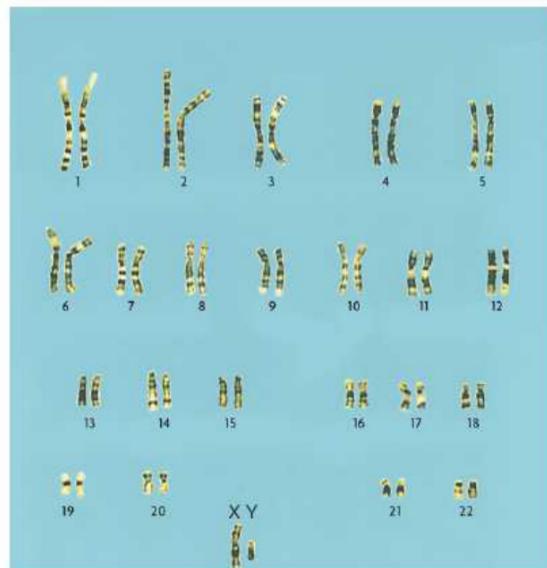
- If the phenotype is more common in males, the gene is likely sex-linked.



## 7.4 Human Genetics and Pedigrees

- Several methods help map human chromosomes.

- A karyotype is a picture of all chromosomes in a cell.



## 7.4 Human Genetics and Pedigrees

- Karyotypes can show changes in chromosomes.
  - deletion of part of a chromosome or loss of a chromosome
  - large changes in chromosomes
  - extra chromosomes or duplication of part of a chromosome

