CHAPTER 12

GENETICS

Topics Discussed in this chapter

Cell Division

- Sexual and asexual reproduction
- Binary fission
- Eukaryotic Cell Cycle
- Chromatin and chromosomes

Mitosis and Meiosis

- Phases of mitosis
- Phases of meiosis
- Tetrads, synapsis and crossing over
- Somatic cells and sex cells
- Autosomes and sex chromosomes

CELL DIVISION

and

REPRODUCTION

Methods of Reproduction

- **Living organisms reproduce by two methods**

1. Asexual reproduction

- **Offspring are identical to the original cell or organism**
- **Involves inheritance of all genes from one parent**
- **Prokaryotes reproduce asexually by binary fission.**

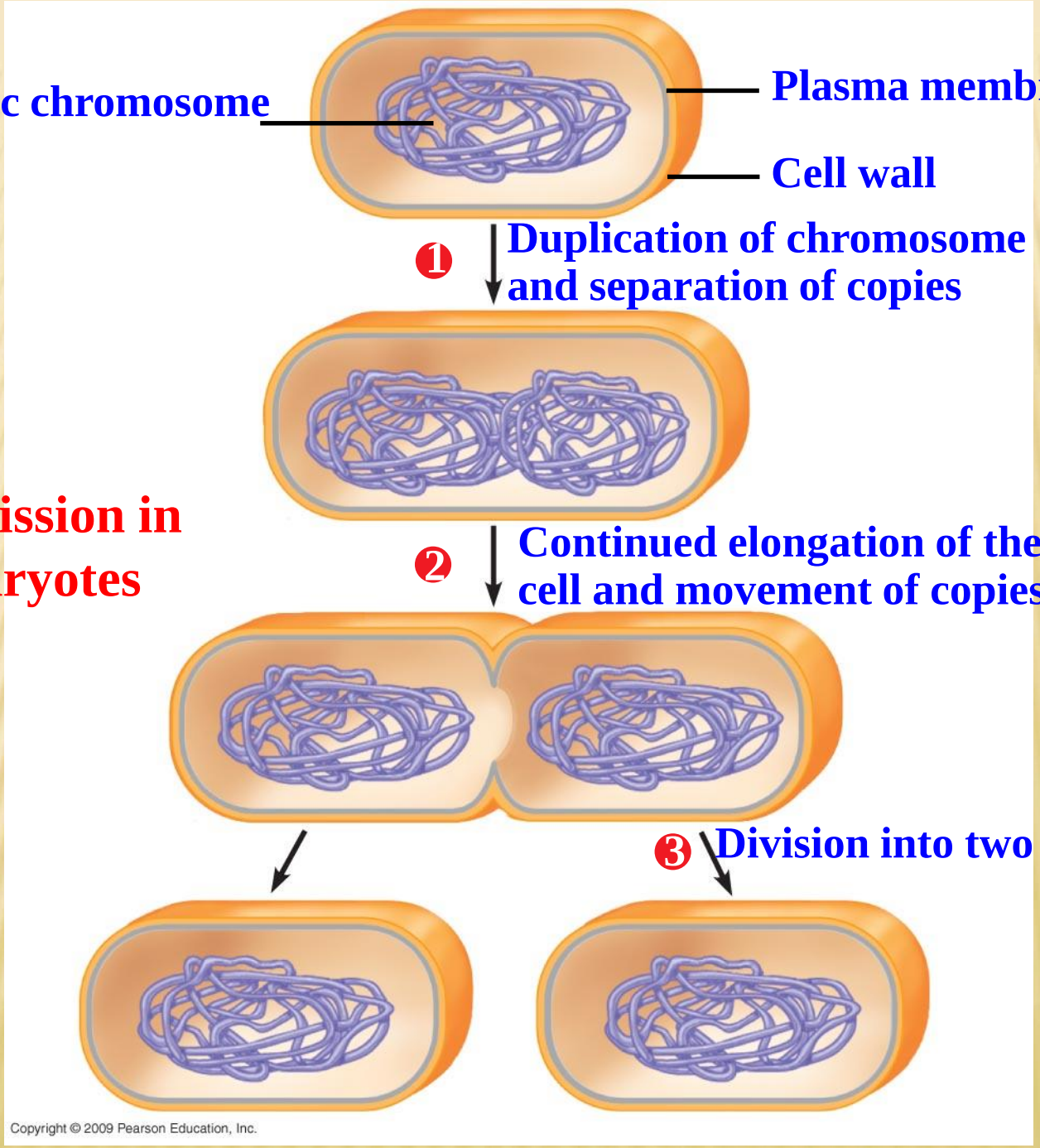
2. sexual reproduction

- **Involves inheritance of unique sets of genes from two parents**
- **Offspring are similar to parents, but show variations in traits**

Prokaryotes reproduce by binary fission

- **Binary fission means "dividing in half"**
  - **Occurs in prokaryotic cells**
  - **Two identical cells arise from one cell**
  - **Steps in the process:**
    - **A single circular chromosome duplicates, and the copies begin to separate from each other**
    - **The cell elongates, and the chromosomal copies separate further**
    - **The plasma membrane grows inward at the midpoint to divide the cells**

Binary fission in Prokaryotes



Eukaryotic Cell Division and Cell Cycle

The cell cycle is an ordered sequence of events for cell division.

- Cells divide when they reach a certain size.
- The cell cycle consists of two stages

1. **Interphase:** Includes G1, S, and G2 phases during which cell contents are duplication .

   G1: first gap phase, growth and prepares for S-phase

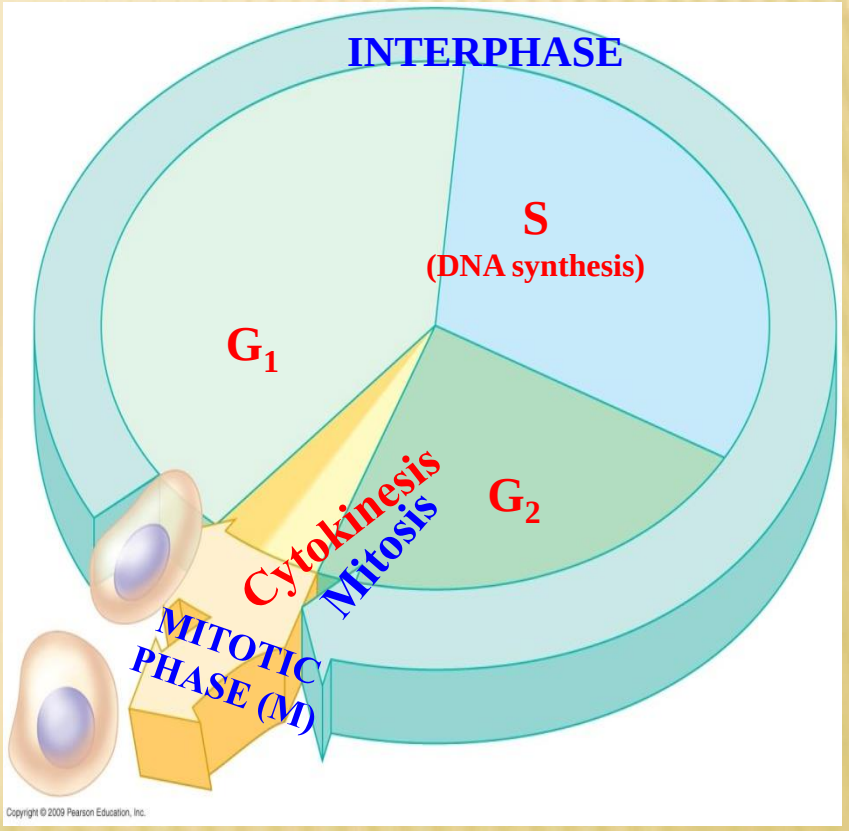
   S: DNA synthesis phase, duplication of chromosomes, each becomes two sister chromatids

   G2: second gap phase, growth and preparation for division

2. **Mitotic phase:** (the M phase) involves mitosis and cytokinesis.

Mitosis: division of the chromosomes

Cytokinesis: division of cytoplasm



The eukaryotic cell cycle

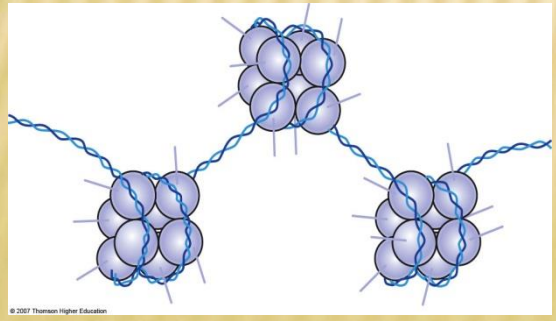
Eukaryotic chromosomes

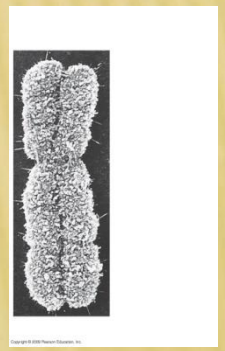
- The chromosomes carry the genetic information.
- Eukaryotic chromosomes contain DNA and protein
- The chromosomes are so named because they may be stained by certain dyes
- When cells are not dividing, the genetic material is decondensed and is called chromatin
- When cells are dividing, the genetic material is condensed and is called chromosome

condensed

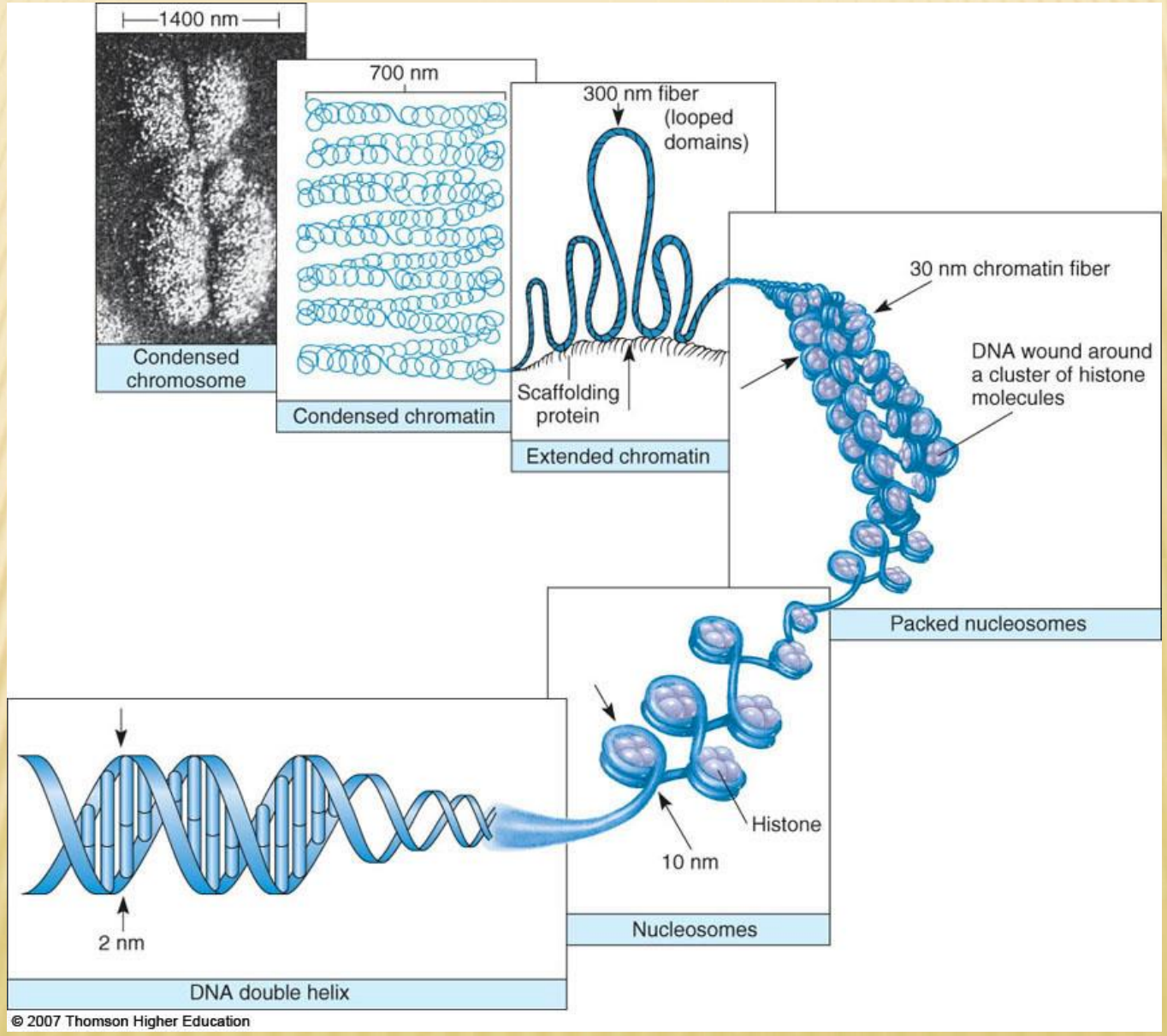
Chromatin ⇌ chromosome

decondensed

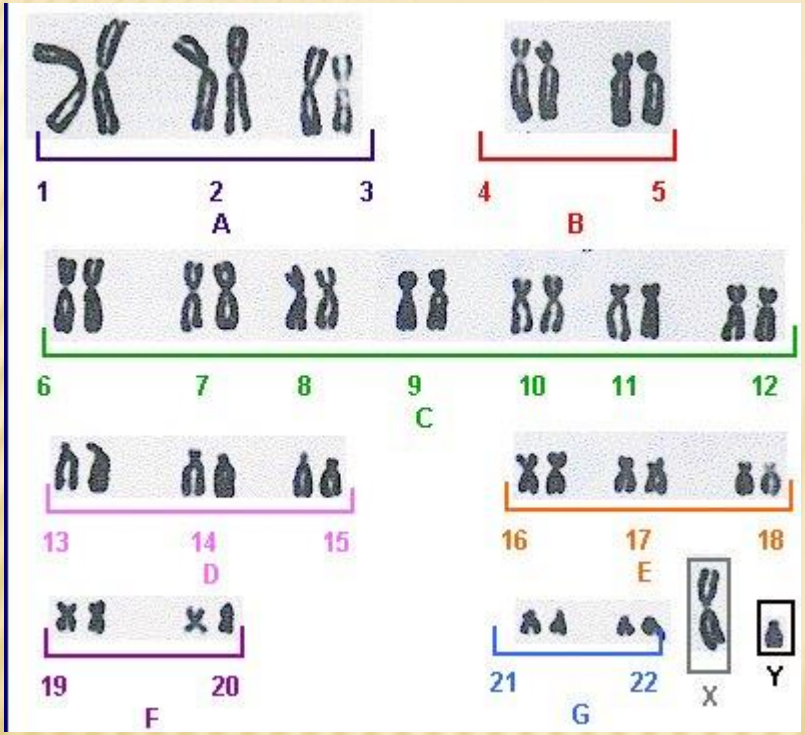




Chromosome Organization



Chromosomes, Mitosis and Meiosis



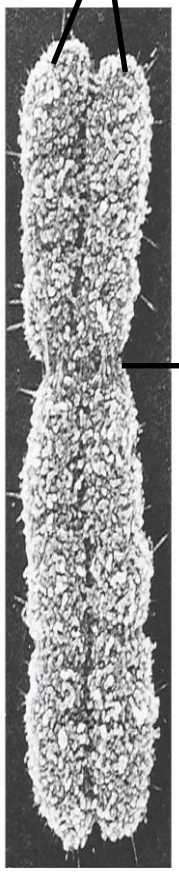
Human chromosomes karyotype

Human chromosomes metaphase spread

The large, complex chromosomes of eukaryotes duplicate with each cell division

- Early in the division process, chromosomes duplicate in S-phase.
- Each chromosome appears as two sister chromatids containing identical DNA molecules.
- Sister chromatids are joined at a narrow region called the centromere.

A Duplicated Chromosome



Sister chromatids

Centromere

Electron micrograph
of a duplicated chromosome

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Chromosome duplication

Sister chromatids

Chromosome distribution To daughter cells

Chromosome duplication and distribution

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Mitosis

- **Identical chromosomes are distributed to each daughter cell**
- **Mitosis preserves chromosome number in eukaryotic cell**

Stages of Mitosis

- **Mitosis: progresses through a series of stages:**

1.Prophase:Chromatin condenses into duplicated chromosomes (pair of sister chromatids) and chromosomes become visible.

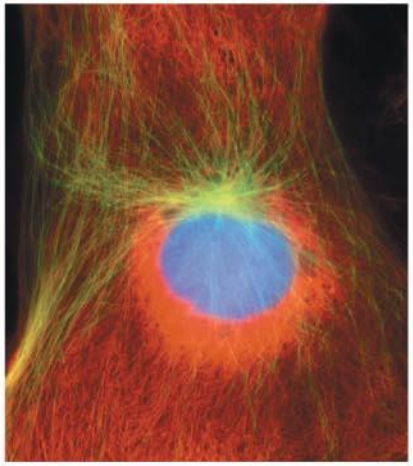
2.Prometaphase:Chromosomes begin to move toward cell's midplan.

3. Metaphase: Chromosomes align on cell's midplane on top of each other.

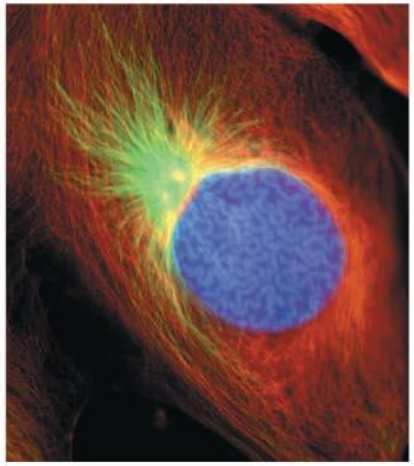
4.Anaphase: Sister chromatids separate, move to opposite poles. Each former chromatid is now a chromosome.

5.Telophase: Chromosomes decondensed. Cytokinesis begins

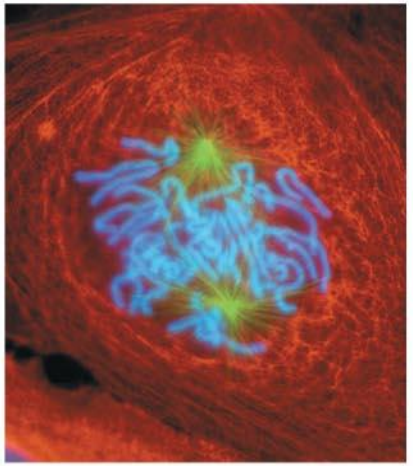
Cytokinesis: Cytoplasmic division. Often overlaps telophase



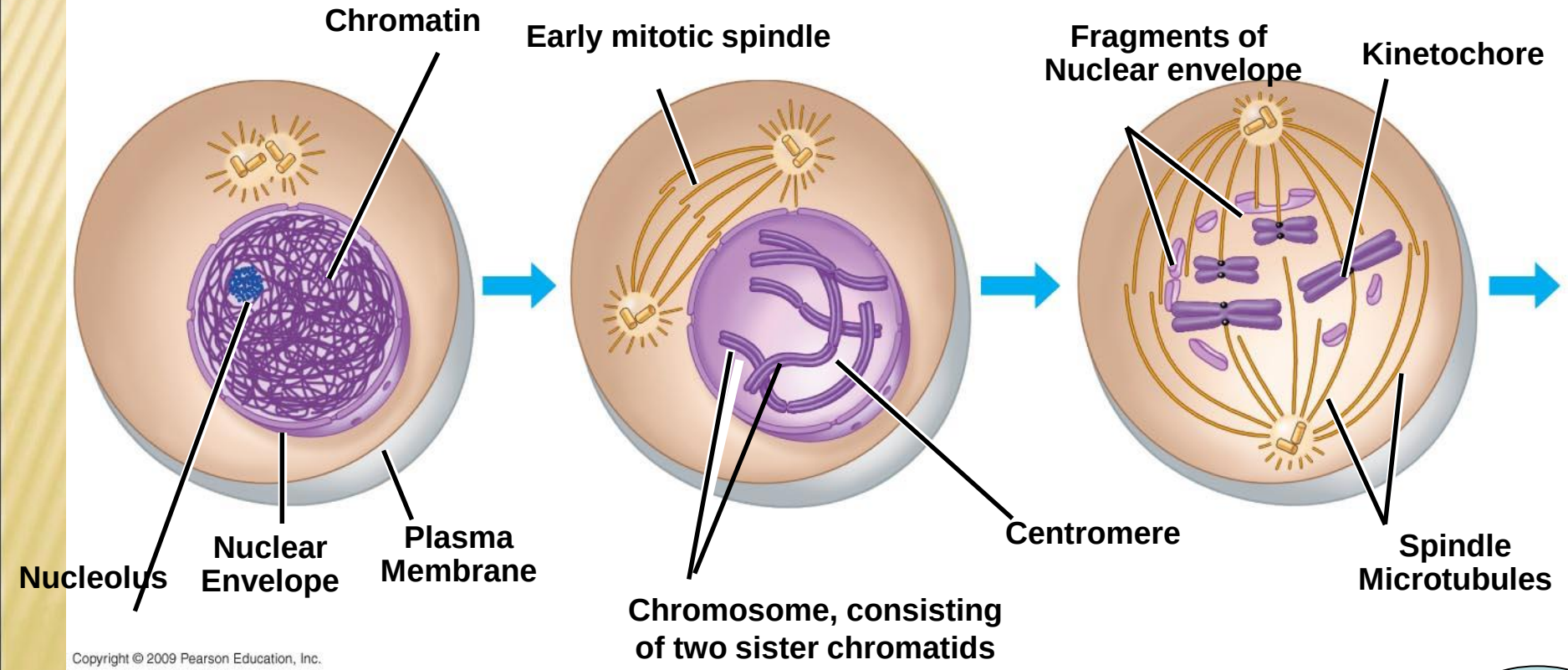
INTERPHASE



PROPHASE

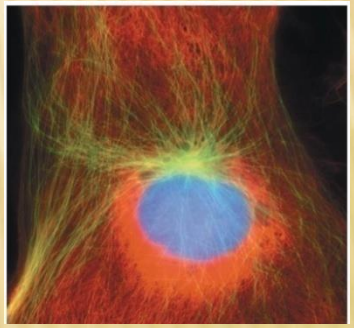


PROMETAPHASE



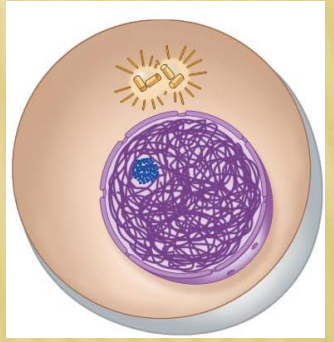
Cell division is a continuum of dynamic changes

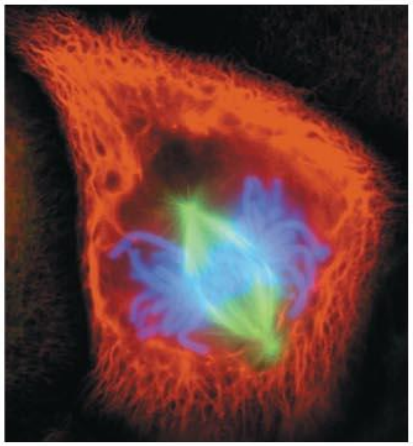
- **Interphase**

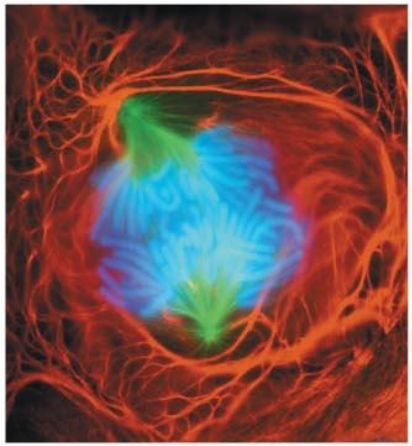


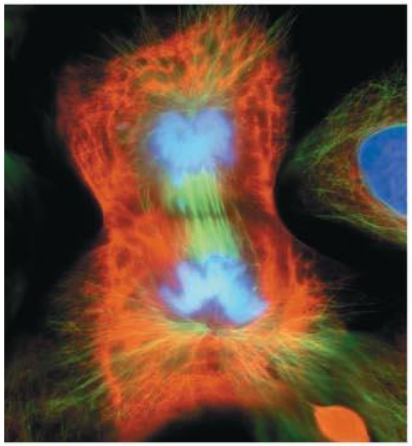
  - **In the cytoplasm**
    - **Cytoplasmic contents double**
  - **In the nucleus**

    Chromosomes duplicate during the S phase





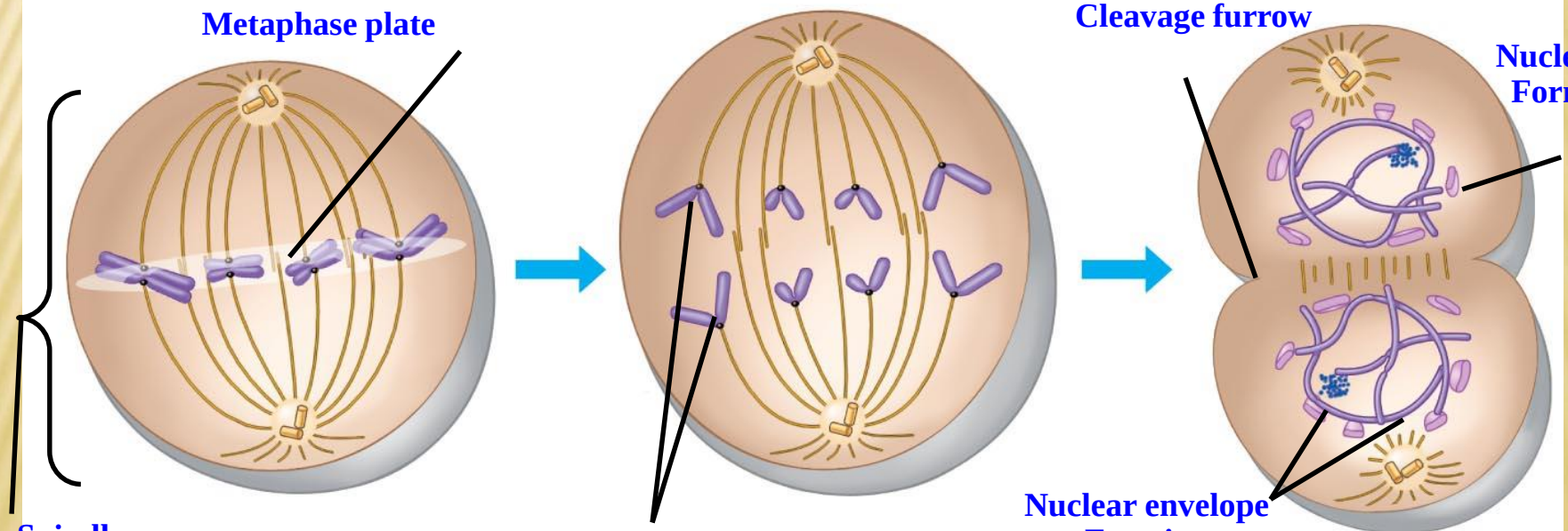




METAPHASE

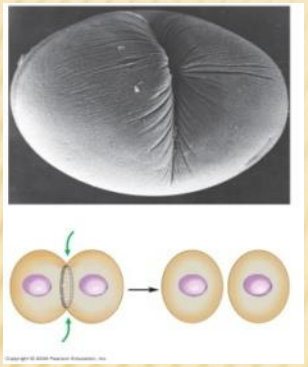
ANAPHASE

TELOPHASE AND CYTOKINESIS



Cytokinesis differs for plant and animal cells

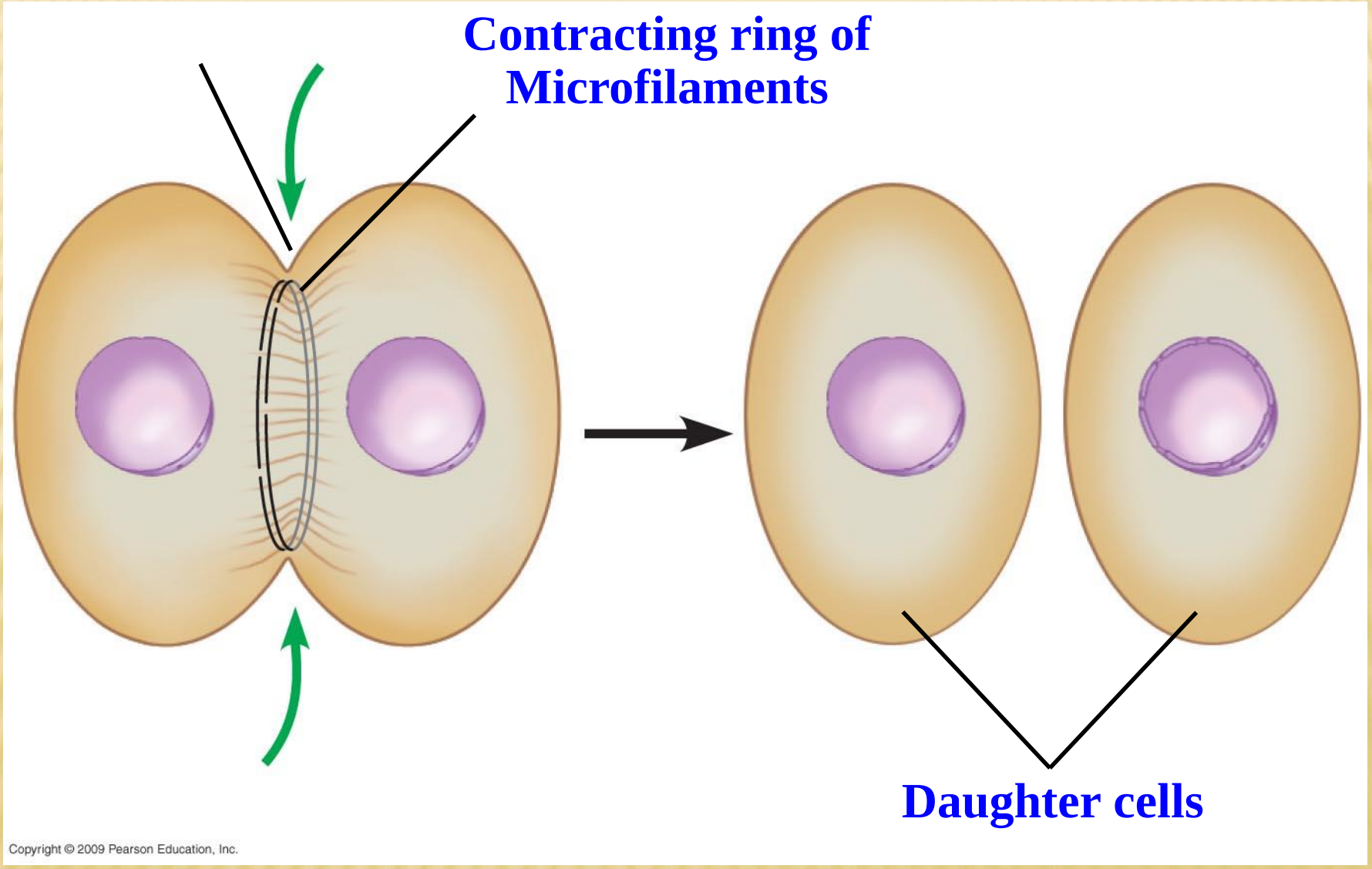
- **Cytokinesis**



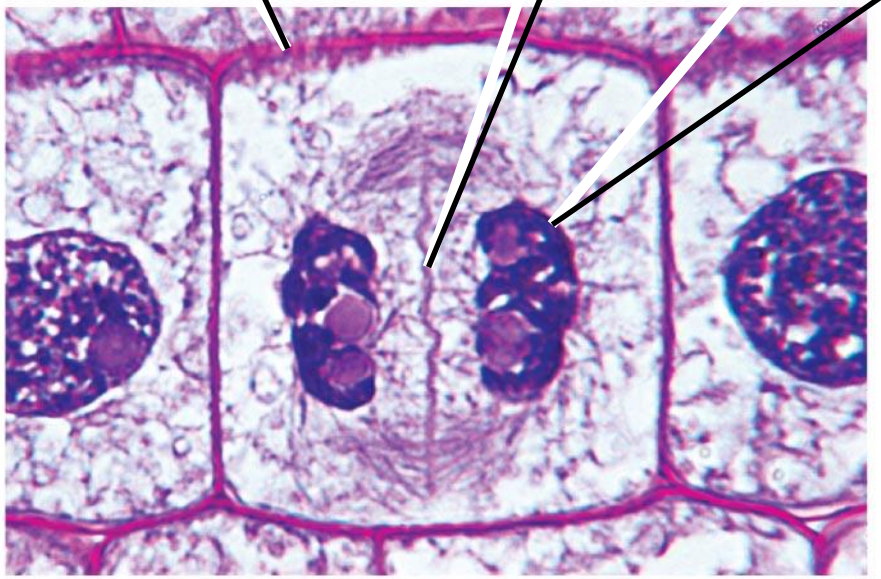
  - **Cleavage in animal cells**
    - **A cleavage furrow forms from a contracting ring of microfilaments, interacting with myosin**
    - **The cleavage furrow deepens to separate the contents into two cells**

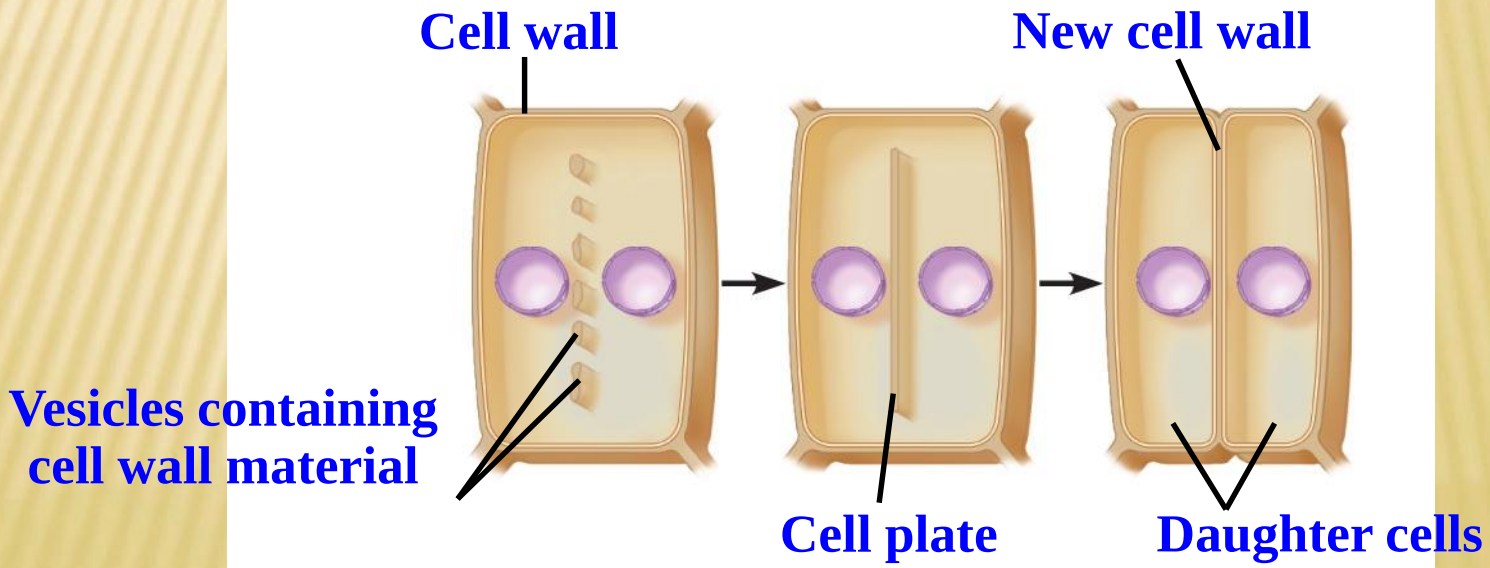
  - **Cytokinesis in plant cells**
    - **A cell plate forms in the middle from vesicles containing cell wall material**
    - **The cell plate grows outward to reach the edges, dividing the contents into two cells**
    - **Each cell has a plasma membrane and cell wall**

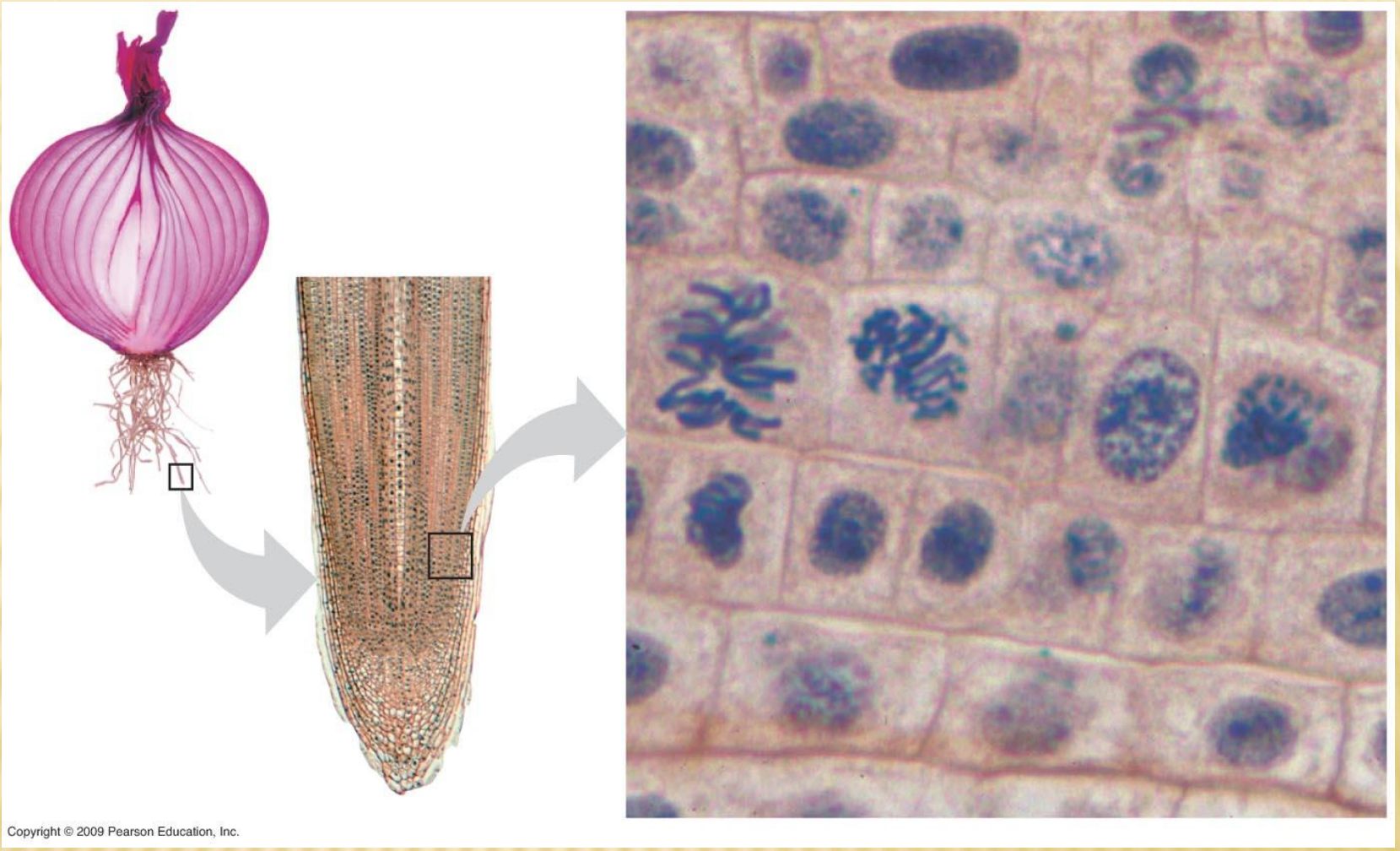
Cytokinesis in animal cells



Cytokinesis in plant cells

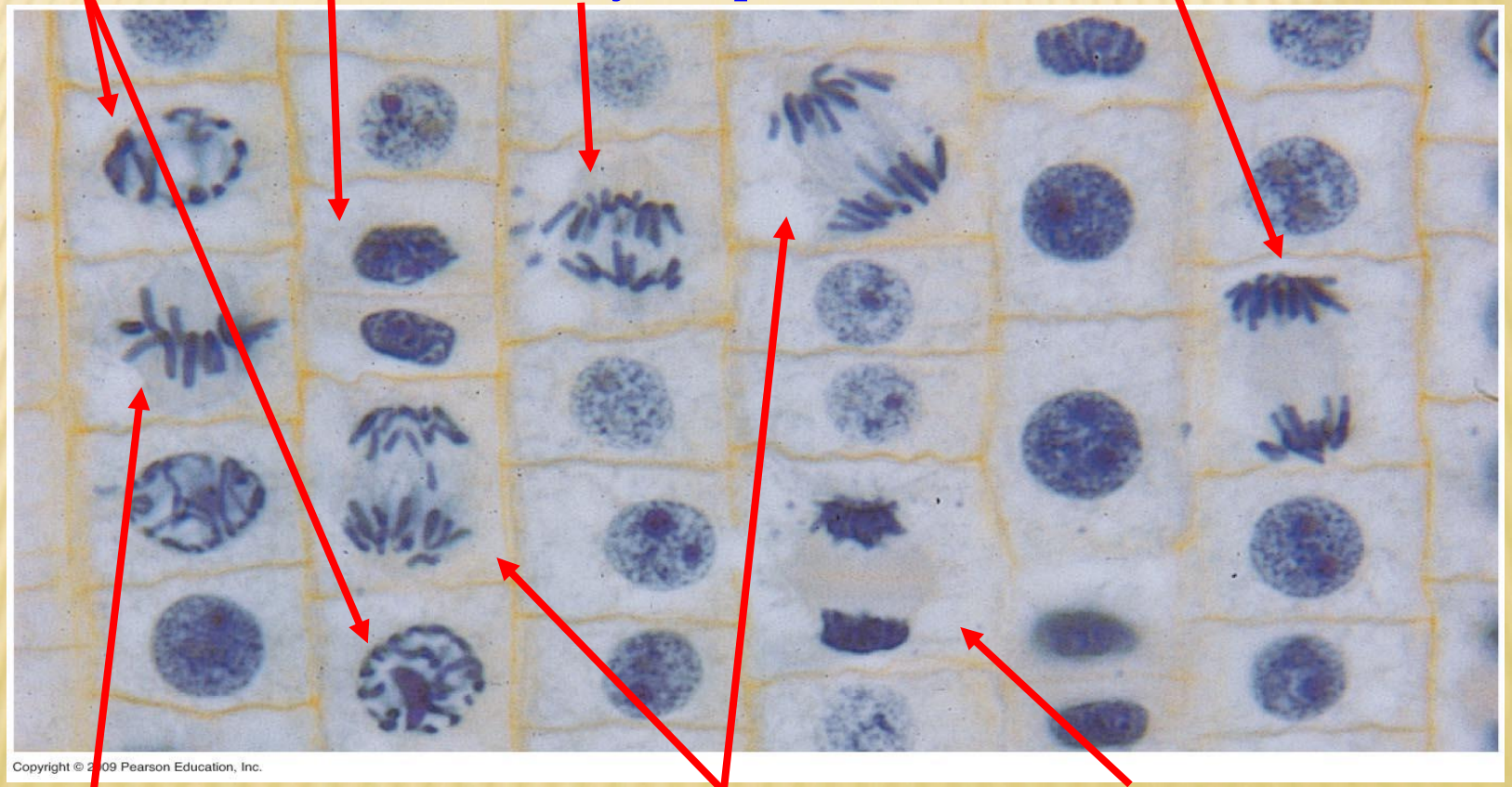






Growth (in an onion root)

Mitosis


prophase
Telophase
Early Anaphase
Late Anaphase
Metaphase
Midi Anaphase
Telophase
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MEIOSIS

Chromosomes are matched in homologous pairs

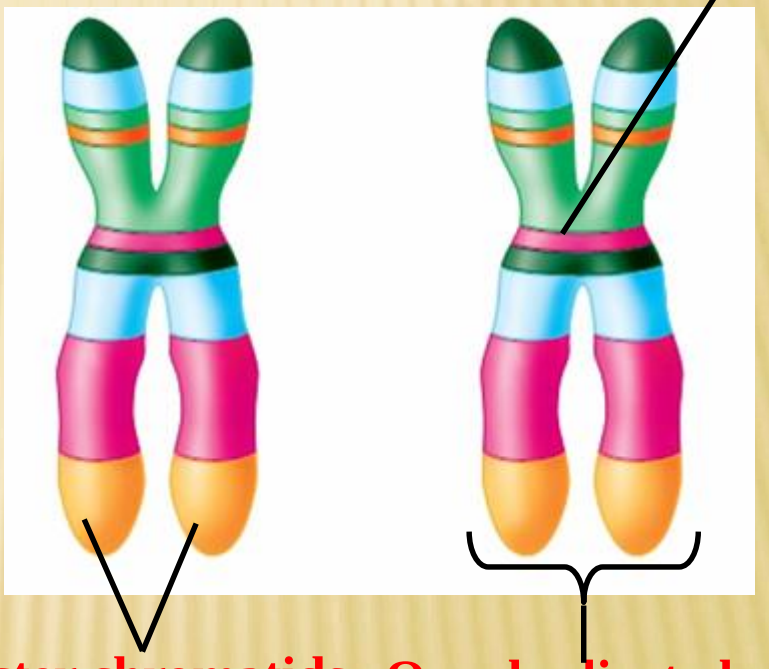
- Somatic cells (all body cells except sex cells, sperm and ovum) have pairs of homologous chromosomes, receiving one member of each pair from the father and one from the mother

- Homologous chromosomes are matched in
  - Length
  - Centromere position
  - Gene locations
    - A locus (plural, *loci*) is the position of a gene
    - Different versions of a gene may be found at the same locus on maternal (mother) and paternal (father) chromosomes

Chromosomes are matched in homologous pairs

Homologous pair of chromosomes

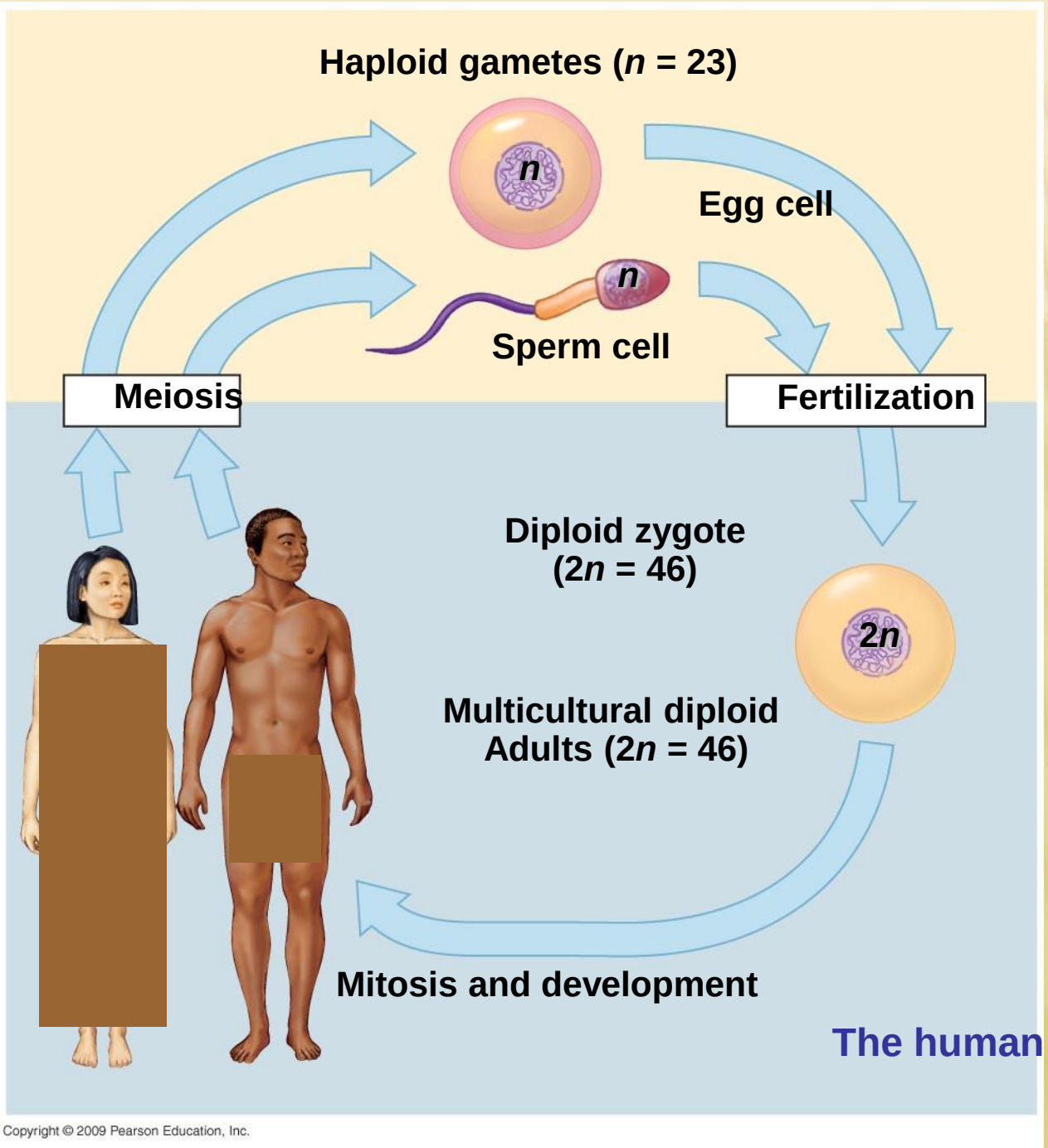
- The human sex chromosomes X and Y differ in size and genetic composition
- Pairs of autosomes (all chromosomes other than sex chromosomes, X &Y) have the same size and genetic composition



A homologous pair of chromosomes

Gametes have a single set of chromosomes

- Meiosis is a process that converts diploid nuclei to haploid nuclei
  - Diploid cells have two homologous sets of chromosomes (2n)
  - Haploid cells have one set of chromosomes (1n)
  - Meiosis occurs in the sex organs (testes and ovaries) producing gametes (sperm and eggs)
- Fertilization is the union of sperm and egg
  - The zygote has a diploid chromosome number, one set from each parent

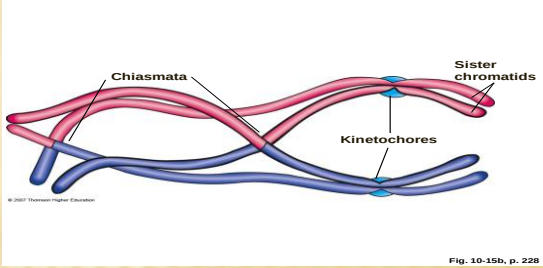


Meiosis reduces the chromosome number from diploid to haploid

- **Like mitosis, meiosis is preceded by interphase**
  - **Chromosomes duplicate during the S-phase**
- **Unlike mitosis, meiosis has two divisions**
  - **During meiosis I, homologous chromosomes separate**
    - **The chromosome number is reduced by half**

      $2n \rightarrow 1n$
  - **During meiosis II, sister chromatids separate**
    - **The chromosome number remains the same $1n$**

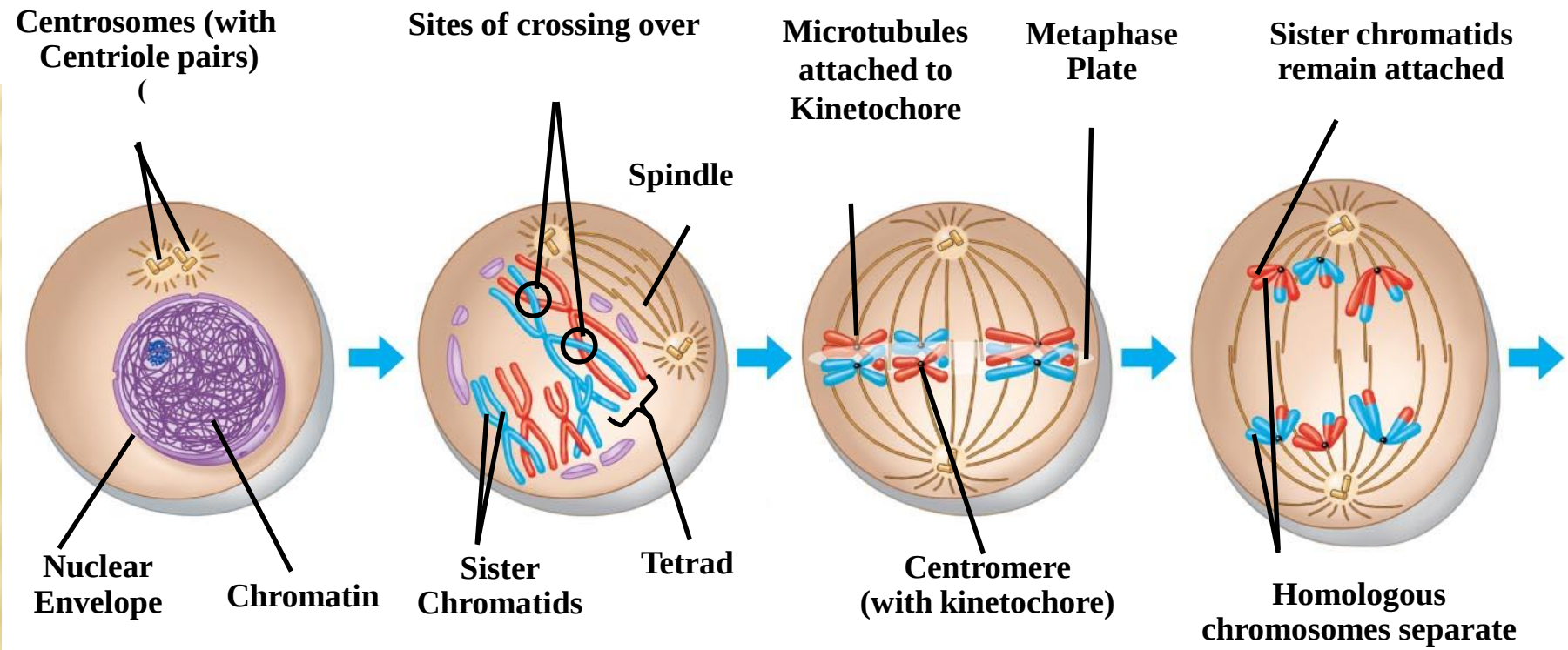
Meiosis reduces the chromosome number from diploid to haploid



- **Events in the nucleus during meiosis I**
  - **Prophase I**
    - **Chromosomes coil and become compact**
    - **Homologous chromosomes come together as pairs by synapsis**
    - **Each pair, with four chromatids, is called a tetrad**
    - **Nonsister chromatids exchange genetic materials by crossing over**
  - **Metaphase I**
    - **tetrads (duplicated homologous chromosomes) line up on metaphase plate side by side**
  - **Anaphase I**
    - **homologous chromosomes separate distributed to different nuclei**
    - **Each nucleus contains haploid number of chromosomes**
    - **Each chromosome has 2 chromatids**
  - **Telophase I and cytokinesis**

MEIOSIS I: Homologous chromosomes separate

INTERPHASE	PROPHASE I	METAPHASE I	ANAPHASE I



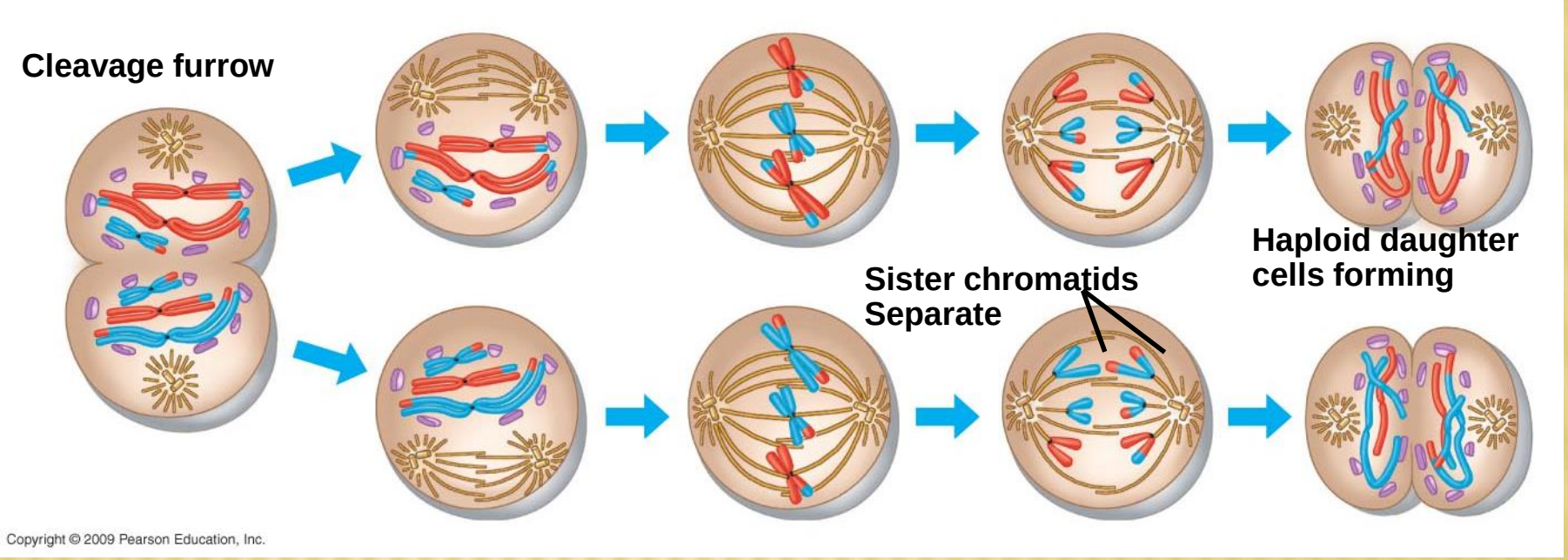
The stages of miosis I

Meiosis II

- Sister chromatids of each chromosome separate
  - one distributed to each daughter cell
- Each former chromatid is now called a chromosome

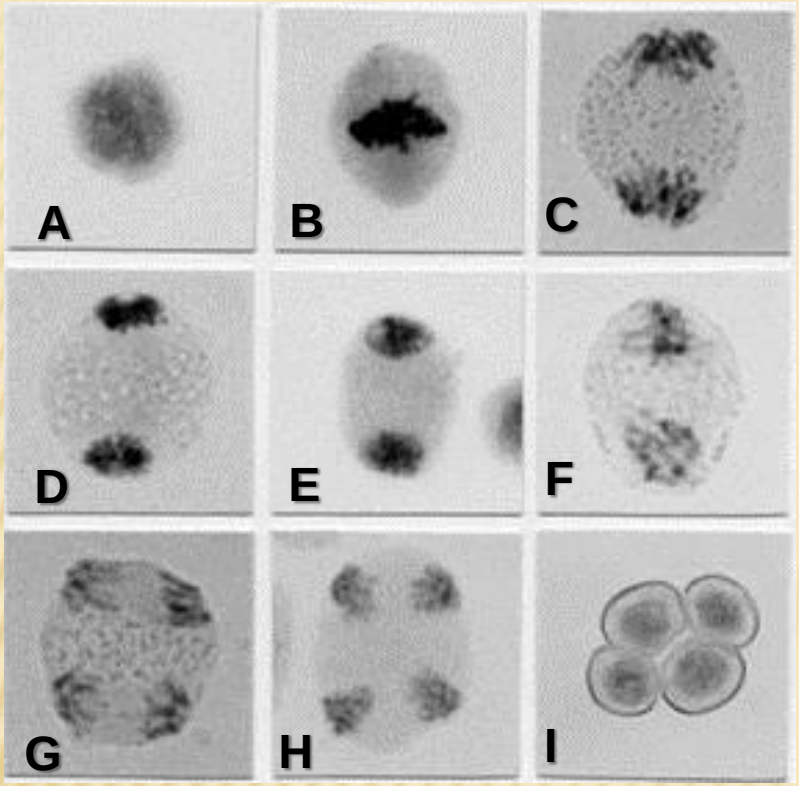
MEIOSIS II: Sister chromatids separate

TELOPHASE I AND CYTOKINESIS	PROPHASE II	METAPHASE II	ANAPHASE II	TELOPHASE II AND CYTOKINESIS

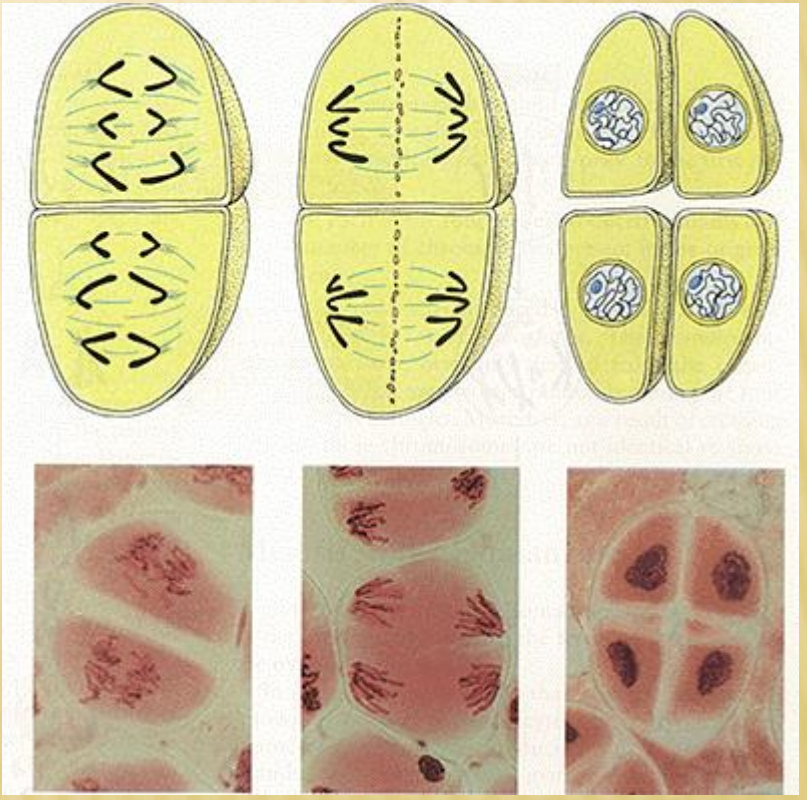


The stages of miosis II

MEIOSIS



A. PROPHASE I
B. METAPHASE I
C. ANAPHASE I
D. TELOPHASE I
E. PROPHASE II
F. METAPHASE II
G. ANAPHASE II
H. TELOPHASE II
I. TETRAD



ANAPHASE II TELOPHASE II TETRAD

Patterns of Inheritance

Topics Discussed in this chapter

- Mendel's laws
  - Mendel's monohybrid pea crosses.
    - True breeding
    - phenotype, genotype
    - Gene, locus, allele
    - dominant allele, recessive allele,
    - homozygous, heterozygous
  - A pedigree
  - Exceptions to Mendel's laws
    - Incomplete dominance, co-dominance
    - Multiple alleles, polygene
    - Pleiotropy
  - Sex determination in different species

MENDEL'S LAWS

Experimental genetics began in a garden

- **Gregor Mendel discovered principles of genetics in experiments with the garden pea**
  - **Mendel showed that parents pass heritable factors to offspring (heritable factors are now called genes)**
  - **Advantages of using pea plants**
    - **Controlled mating**
    - **Self-fertilization or cross-fertilization**
    - **Observable characteristics with two distinct forms**
    - **True-breeding strains**

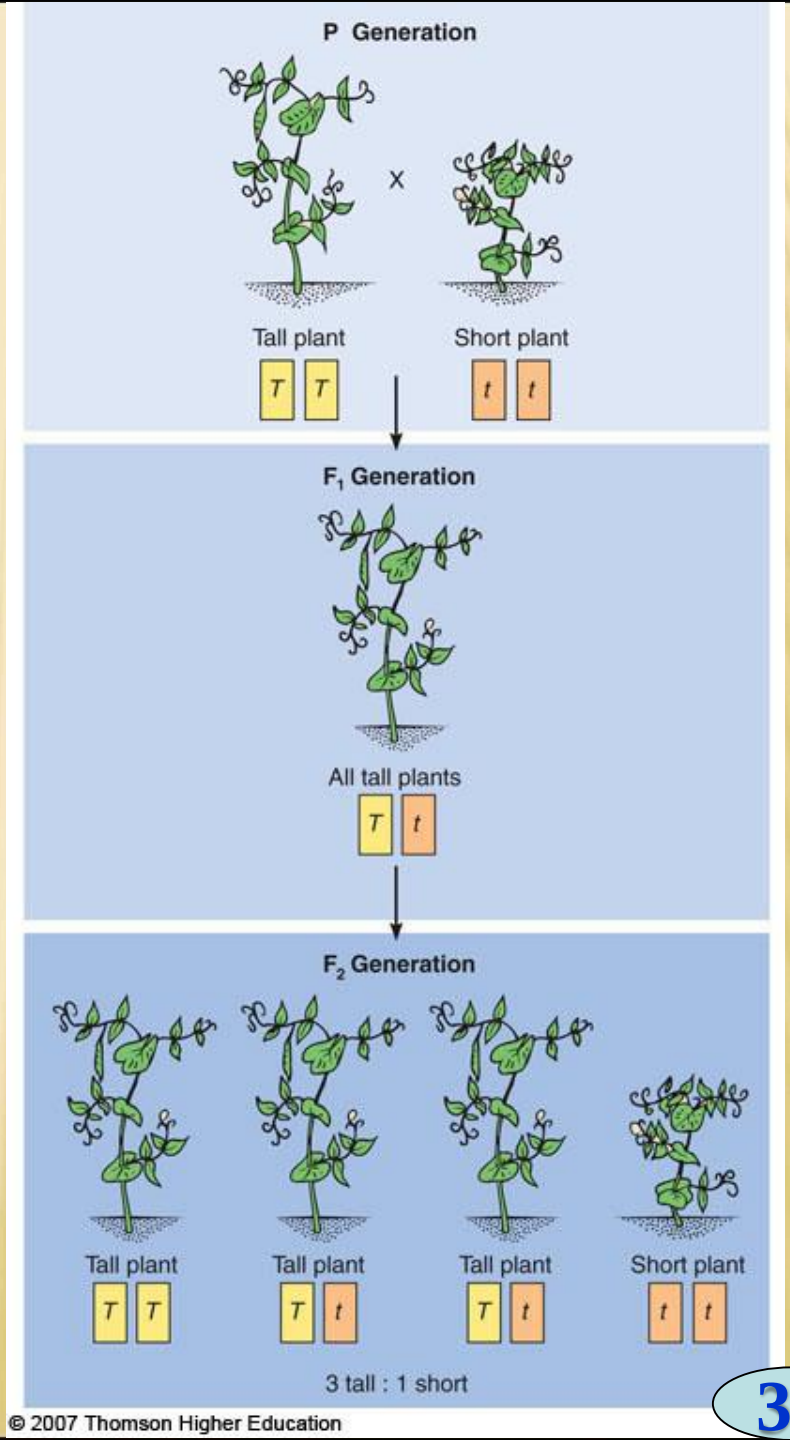
P generation (true-breeding parents)

One of Mendel's pea crosses.

F_1 generation

Fertilization among F_1 plants (F_1 × F_1)

F_2 generation



Mendel's law of segregation describes the inheritance of a single character

- **Example of a monohybrid cross**
  - **Parental generation: Tall plant× Short plant**
  - **F_1 generation: all plants were tall**
  - **F_2 generation: Tall plants and short plants**
- **Mendel needed to explain**
  - **Why one trait seemed to disappear in the F_1 generation**
  - **Why that trait reappeared in one quarter of the F_2 offspring**

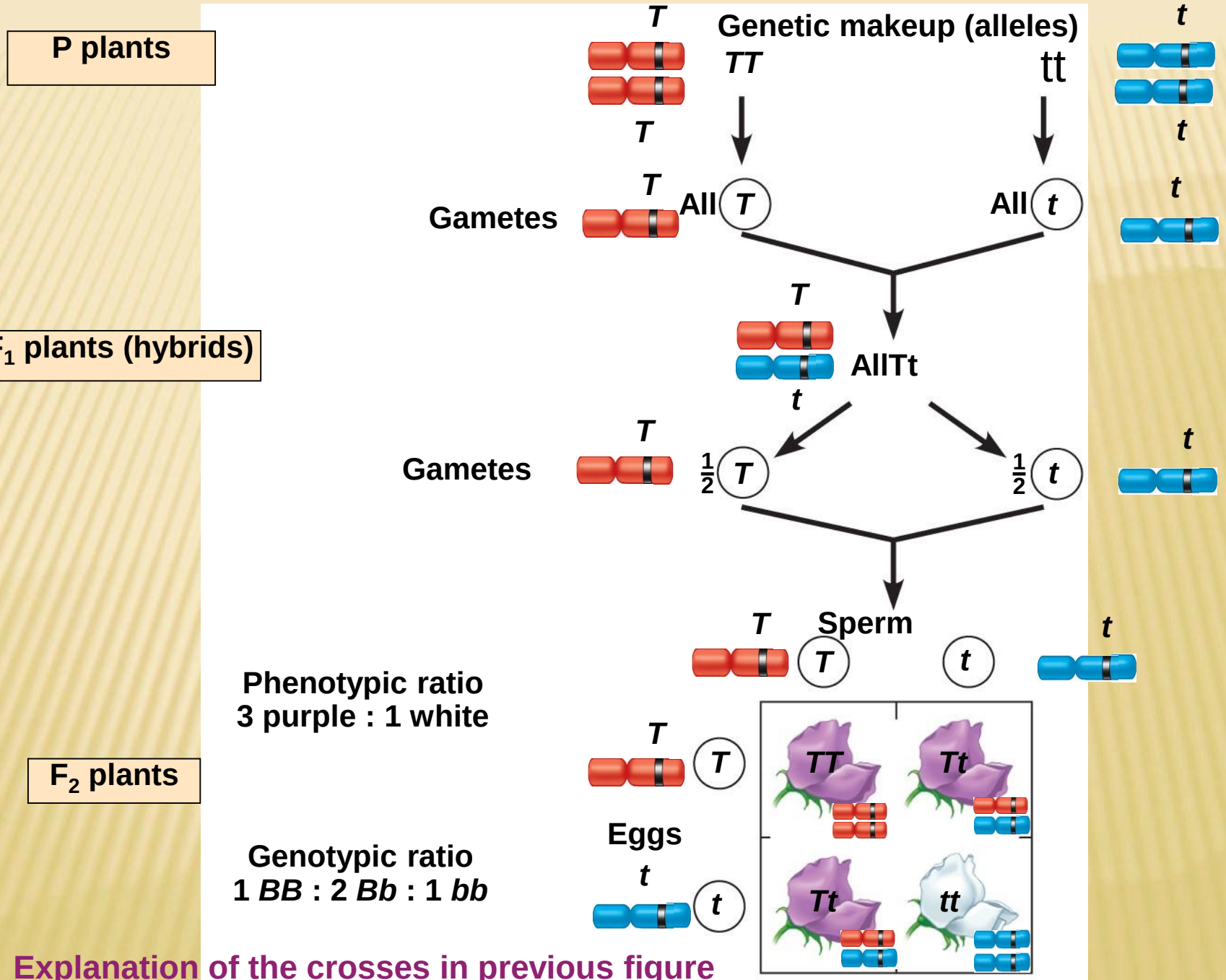
Questions:

Why one trait seemed to disappear in the F_1 generation?

Why that trait reappeared in one quarter of the F_2 offspring?

Answers: The questions were answered by Mendel's Principle of Segregation (separation) which states that:

- **Each trait is controlled by two factors (now known as alleles).**
- **During gametes formation (meiosis) the two alleles segregate (separate), so that each gamete (sperm or ovum) has one allele only.**



Explanation of the crosses in previous figure

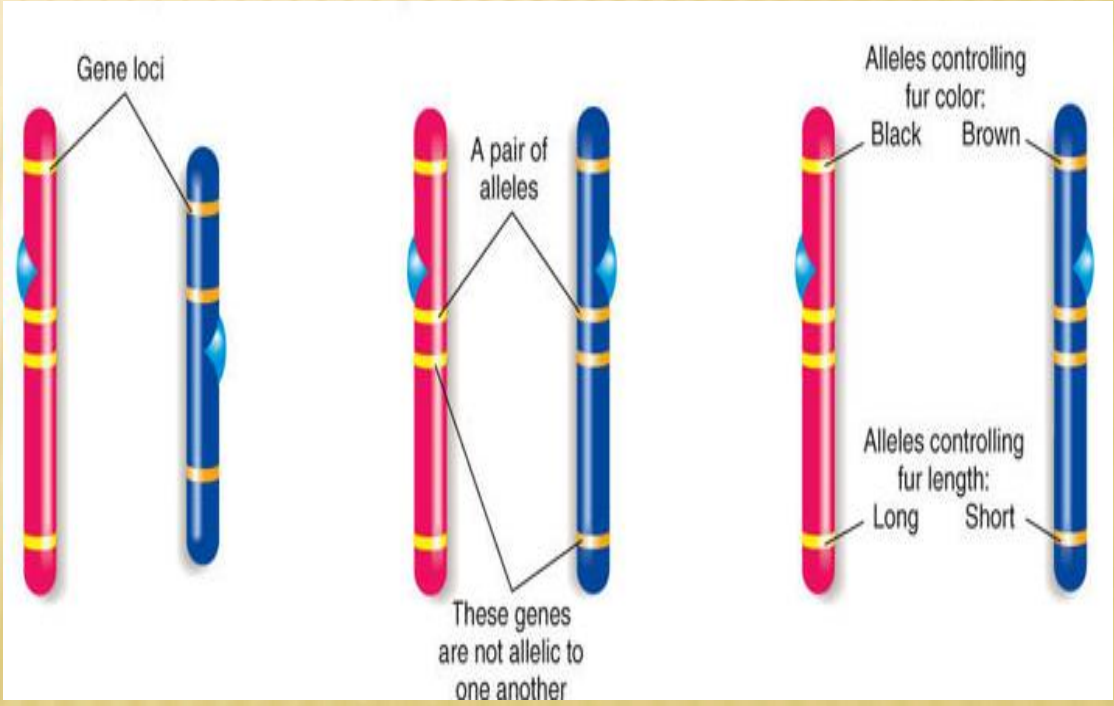
Learning Objective

- **Define the terms**
- **phenotype, genotype**
- **locus, allele**
- **dominant allele, recessive allele**
- **homozygous, and heterozygous**

- **Genes:** information units in chromosomes. There are two copies of each gene. One on the father chromosome and one on the mother chromosome. Each copy is called allele.

- **Locus:** site of a gene on the chromosome.

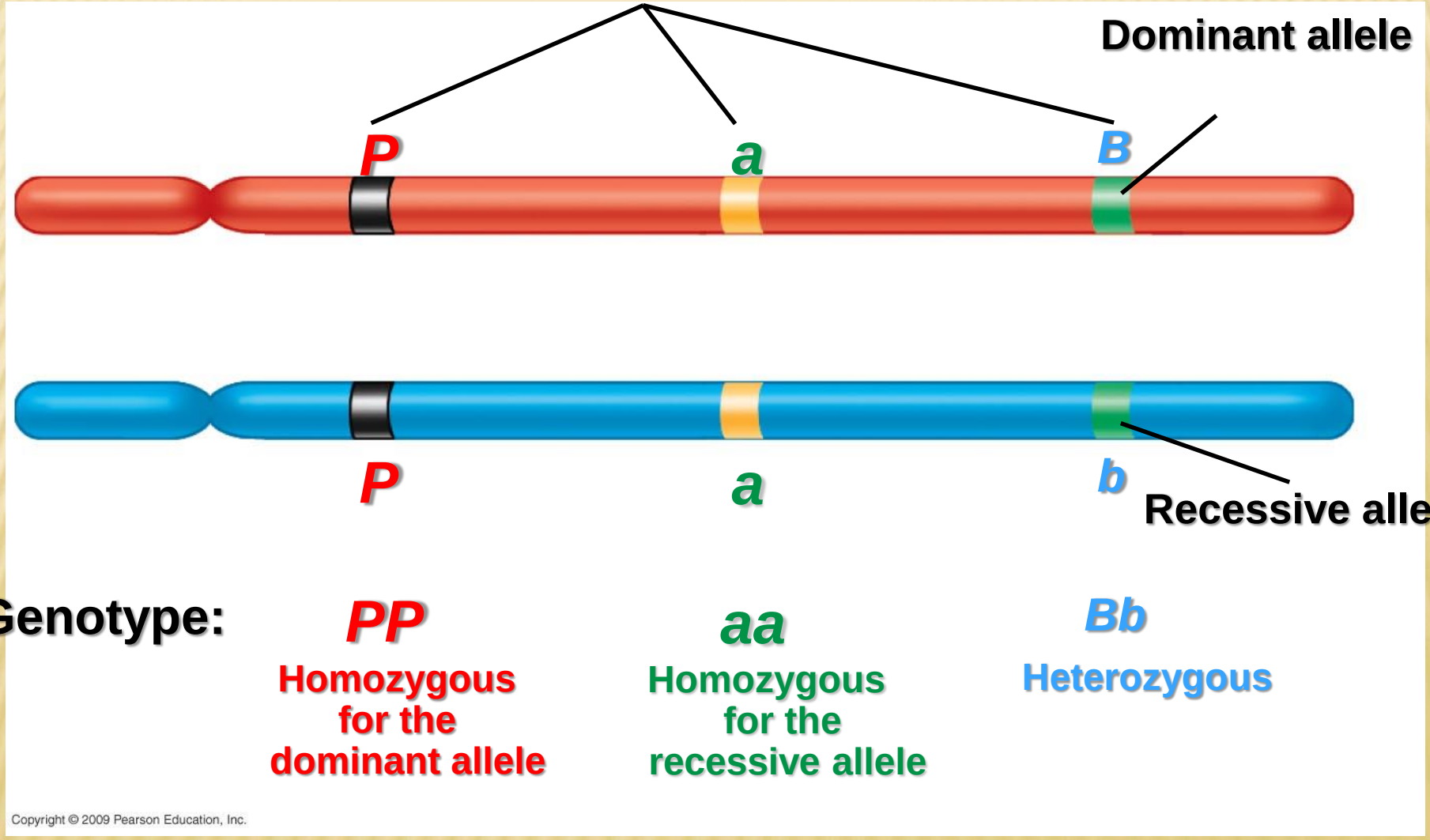
- **Alleles:** Copy of a gene (each gene has 2 copies, one on each of the homologous chromosomes), same loci on homologous chromosomes



Gene Pairs

- **Diploid individuals:** Individual whose cells contain 2 sets of chromosome (23 from the mother egg+23 from the father sperm).
  - Consequently, genes on these homologous chromosomes are in pairs. One from the father and one from the mother. Each copy is called alleles.
- **Homozygous**
  - Two identical alleles e.g. AA or aa.
- **Heterozygous**
  - Two different alleles e.g. Aa.

Gene loci



Genotype:

PP	***aa***	***Bb***
Homozygous for the dominant allele	**Homozygous for the recessive allele**	**Heterozygous**

Matching gene loci on homologous chromosomes

Gene Expression

- **Dominant allele**
  - **Alleles that is expressed in the heterozygous and it masks expression of a recessive allele**
- **Recessive allele**

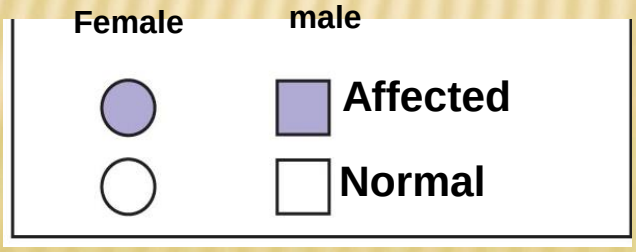
  Alleles that is not expressed in the heterozygous
- **Phenotype**

  appearance
- **Genotype**

  genetic constitution

Genetic traits in humans can be tracked through family pedigrees

- **A pedigree**
  - **Shows the inheritance of a trait in a family through multiple generations**
  - **Can also be used to deduce genotypes of family members.**
  - **Important in genetic counseling.**

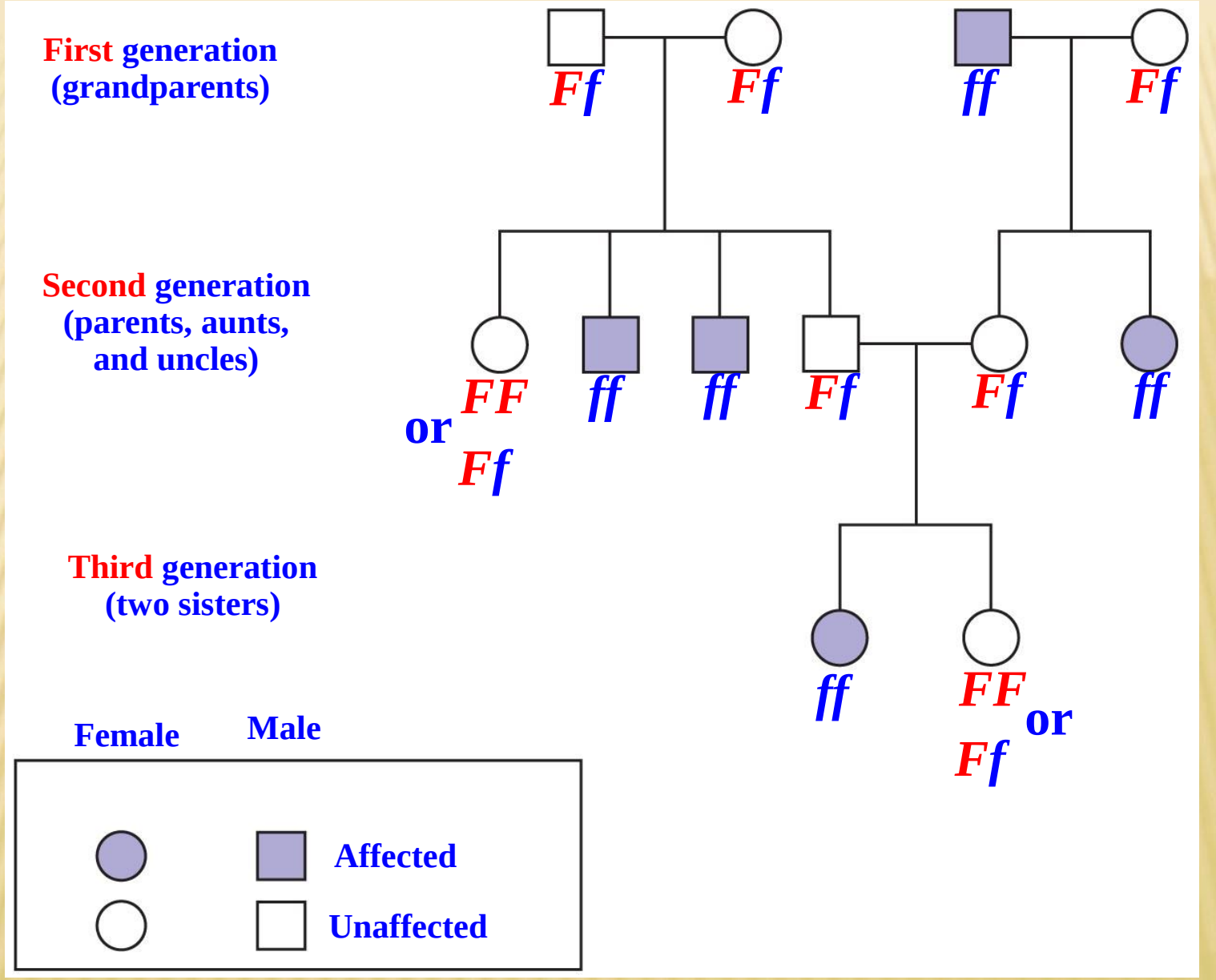


Symbols used in pedigree analysis

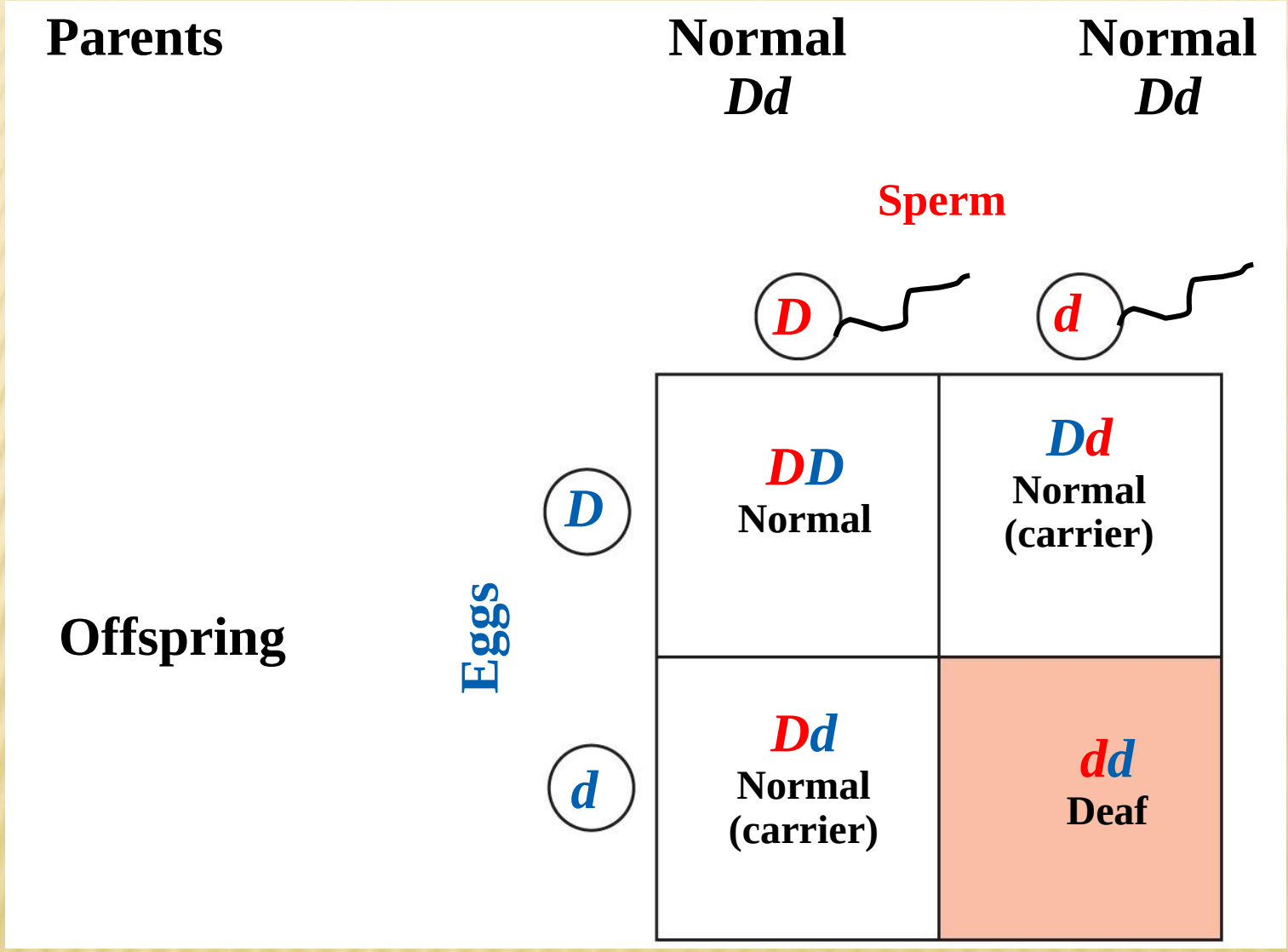
Examples of single-gene inherited traits in humans

Earlobe

	Dominant Traits	Recessive Traits
Genotype	***FF or Ff***	***ff***
		
Phenotype	**Free earlobe**	**Attached earlobe**



Pedigree showing inheritance of attached versus free earlobe in a hypothetical family



Offspring produced by parents who are both carriers for Deafness which is a recessive diorder

Exceptions to Mendel's laws

Variations to Mendel's Laws

Traits inheritance is not always dominant or recessive, or controlled by one gene.

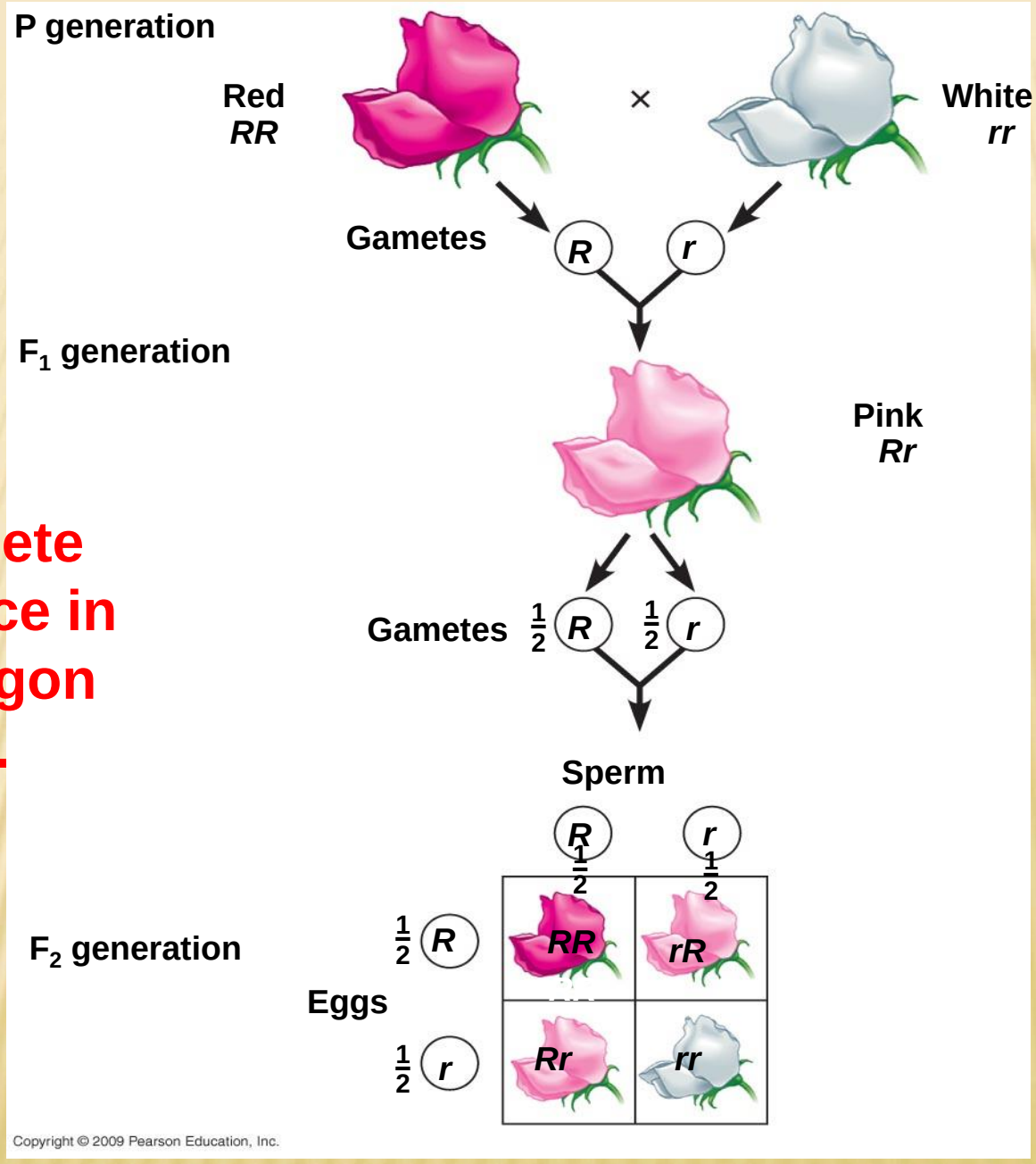
Some of the exceptions to Mendel's Laws are:

1. **Incomplete dominance: heterozygote has intermediate phenotype**
2. **Co-dominance: heterozygote expresses phenotypes of both homozygotes.**
3. **Multiple alleles: Three or more alleles in a population for the same locus. Diploid individual has any two alleles.**
4. **Pleiotropy: the phenomenon of one gene mutation being responsible for or affecting more than one phenotypic characteristic.**
5. **Polygenes: Multiple independent pairs of genes may have similar and additive effects on the phenotype.**

Incomplete dominance results in intermediate phenotypes

- **Incomplete dominance**
  - **Neither allele is dominant over the other**
  - **Expression of both alleles is observed as an intermediate phenotype in the heterozygous individual**

Incomplete dominance in snapdragon color.



Exceptions to to Mendel Laws

When Mendel's laws/results may not be observed

Genetic Occurrence	Definition	Examples
Polygenic inheritance	More than one gene can affect a single trait	• 4 genes are involved in determining eye color. • Human height
Pleiotropy	A single gene can affect more than one trait	• A pleiotropic allele dominant for yellow fur in mice is recessive for lethal developmental defect. •Sickle cell anemia
Multiple alleles for one gene	Genes may have more than two alleles	ABO blood types in humans
Dominance is not always complete	• In incomplete dominance the heterozygote is intermediate. • In co-dominance no single allele is dominant, and the heterozygote shows some aspect of both homozygotes.	• Human blood groups
Environmental factors	Genes may be affected by the environment.	Siamese cats

Sex determination in different species

- X-Y system in mammals, fruit flies
  - XX = female
  - XY = male
- X-O system in grasshoppers and roaches
  - XX = female
  - XO = male
- Z-W in system in birds, butterflies, and some fishes
  - ZW = female
  - ZZ = male
- Chromosome number in ants and bees
  - Diploid = female
  - haploid = male

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Female

♀ 22 + XX

male

♂ 22 + X

X-O system

X-O

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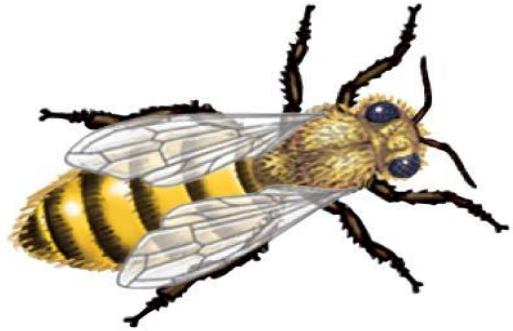
♀ 76 + ZW

♂ 76 + ZZ

Z-W system

Z-W



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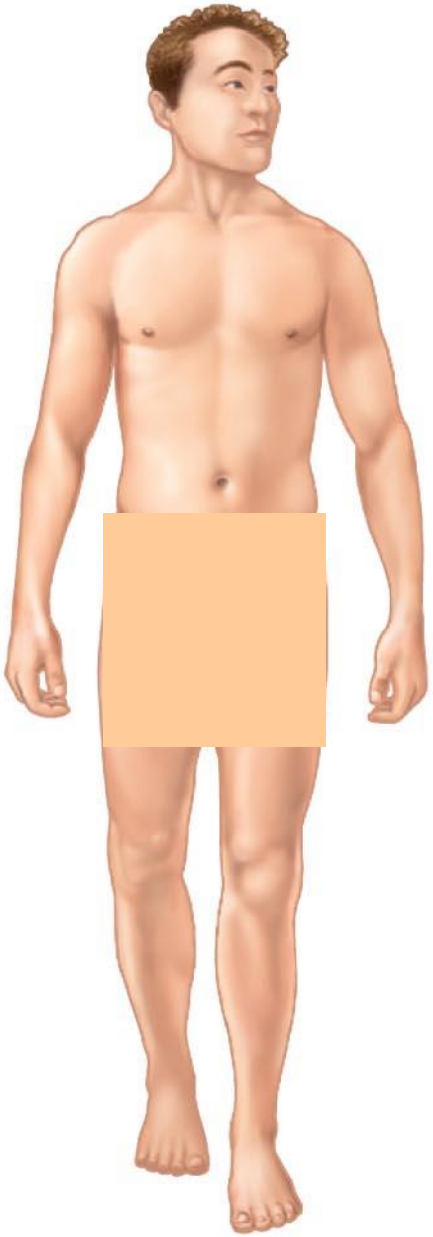
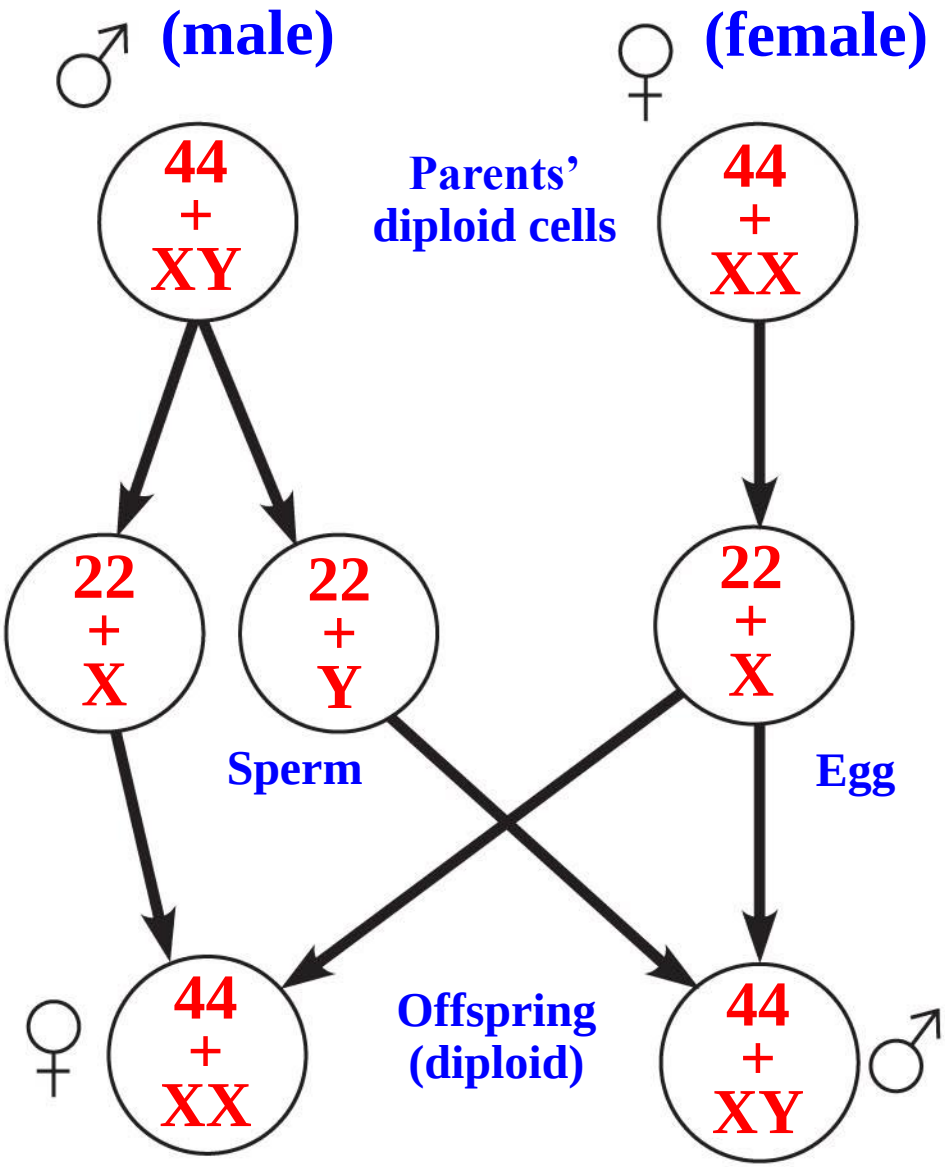
♀ 32

♂ 16

Chromosome number system



X-Y system



Male not female (responsible) for getting either male or female babies

- يقول ✈ تبارك و تعالى فى كتابه الحكيم فى (سورة القيامة):

بسم الله الرحمن الرحيم

أَيَحْسَبُ الْإِنسَانُ أَن يُتْرَكَ سُدًى ﴿36﴾ أَلَمْ يَكُ نُطْفَةً مِّن مَّنِيٍّ يُمْنَى ﴿37﴾ ثُمَّ كَانَ عَلَقَةً فَخَلَقَ فَسَوَّى ﴿38﴾ فَجَعَلَ مِنْهُ الزَّوْجَيْنِ الذَّكَرَ وَالْأُنثَى ﴿39﴾

صدق الله العظيم

- كانت إمرأة أبى حمزة الضبى شاعرة ، و قد هجرها زوجها حين ولدت بنتاً يوماً بخبائها ، فإذا هى تقول:

ما لأبى حمزة لا يأتينا ... يظل فى البيت الذى يـلينا

غضبـان ألا نلد الـبنينا ... تا الله ما ذلك فى ايـــدينا

و إنما نأخذ ما أعطـينا ... و نحن كالأرض لزارعينا

تنبت ما قد زرعوه فينا

فرق لها و صالحها