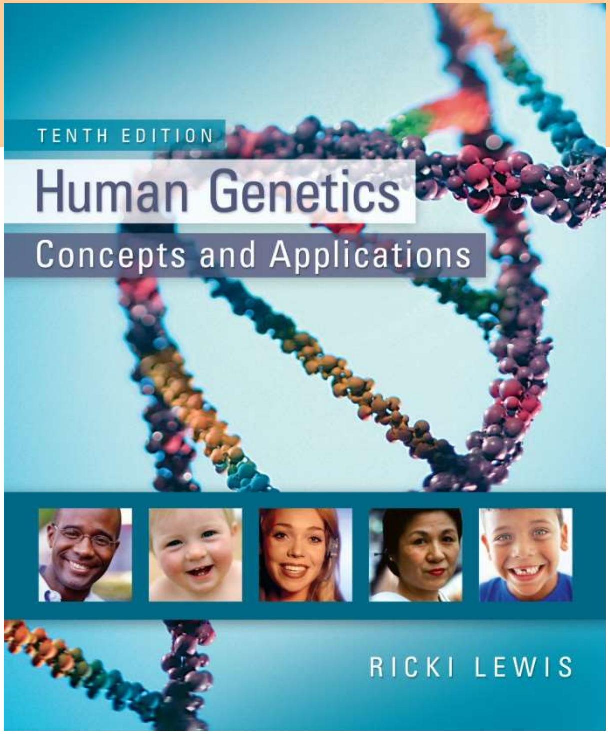
Human Genetics
Concepts and Applications
Tenth Edition

RICKI LEWIS



3 Meiosis and Development

PowerPoint® Lecture Outlines
Prepared by Johnny El-Rady, University of South Florida

Stages of the Human Life Cycle

Genes orchestrate our physiology after conception through adulthood

Development is the process of forming an adult from a single-celled embryo

In humans, new individuals form from the union of sex cells or **gametes**

- **Sperm** from the male and **oocyte** from the female form a **zygote**

The Male Reproductive System

Sperm cells are made in the seminiferous tubules of the **testes**

The prostate gland, seminal vesicles, and bulbourethral glands add secretions to form the seminal fluid

Sperm mature and are stored in the epididymis

They leave through the ductus deferens and then the urethra

The Male Reproductive System

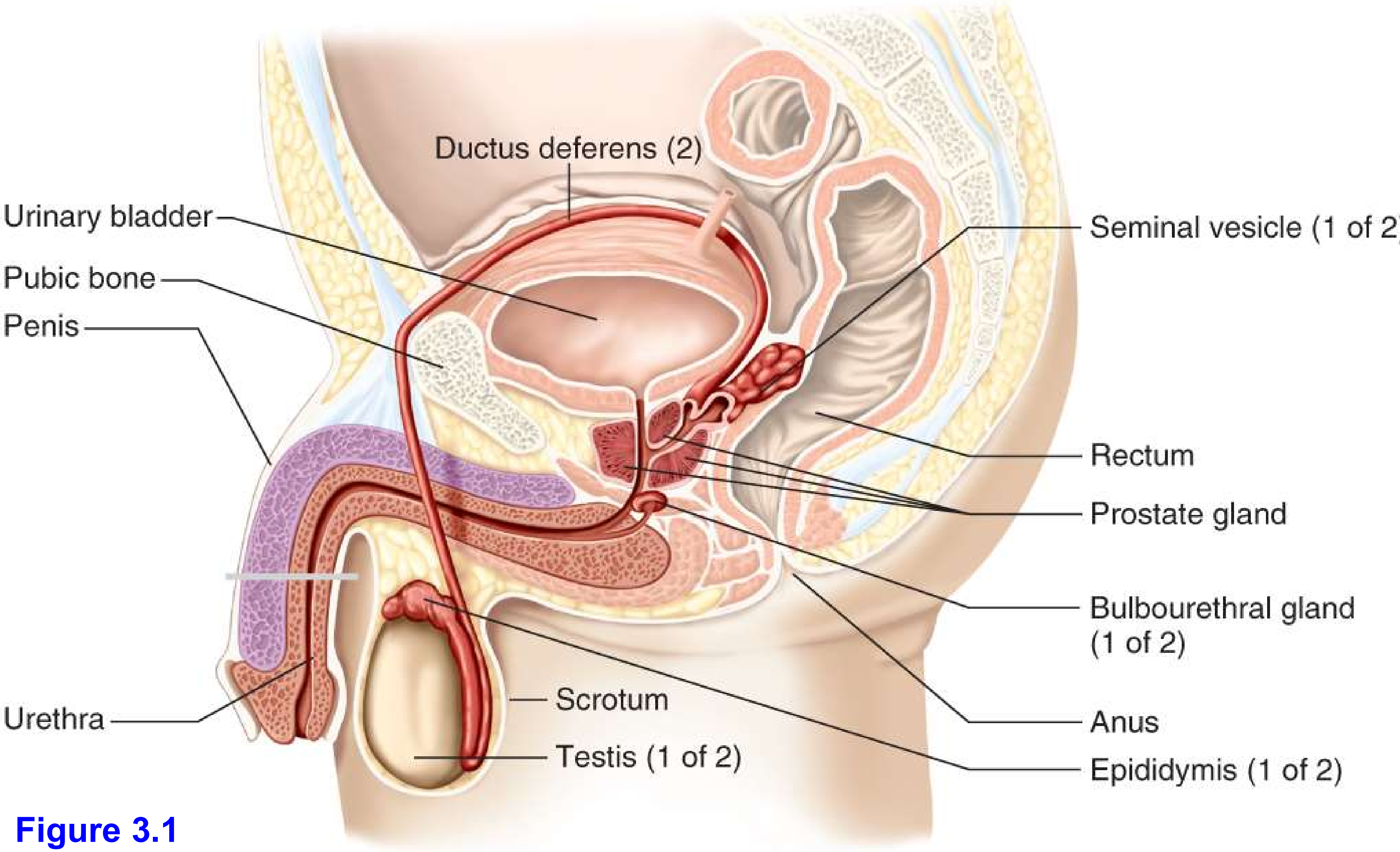


Figure 3.1

The Female Reproductive System

Oocytes mature in the **ovaries**

Each month, an ovary releases an oocyte into the uterine tube

- If the oocyte is fertilized, it continues to the uterus where it divides and develops

- If it is not fertilized, the body expels it, along with the uterine-lining via the menstrual flow

Hormones control the cycle of oocyte development

The Female Reproductive System

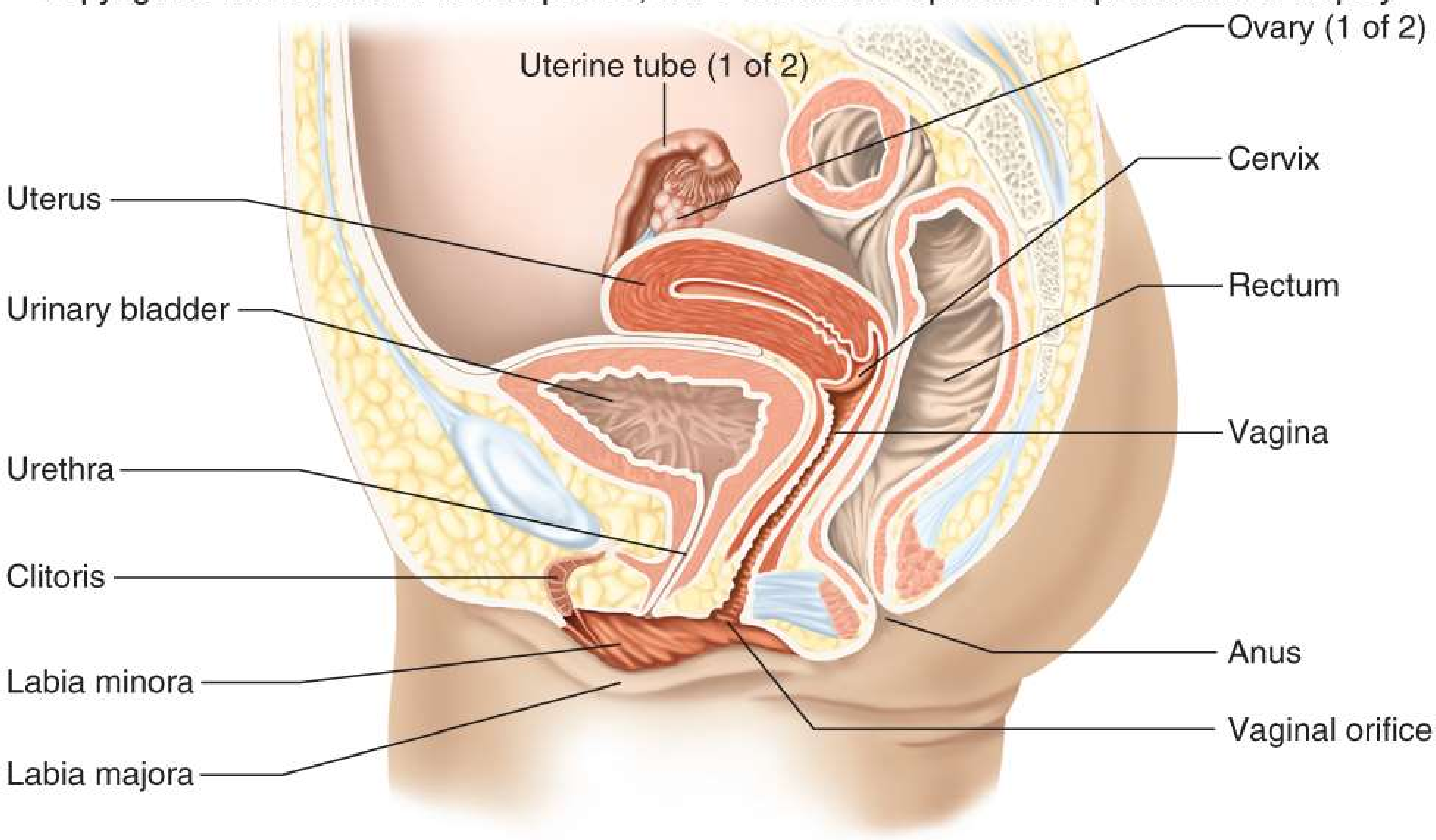


Figure 3.2

Meiosis

The cell division that produces **gametes** with half the number of chromosomes

Occurs in special cells called germline cells

Maintains the chromosome number of a species over generations

Ensures genetic variability via the processes of independent assortment and crossing over of chromosomes

Meiosis consists of two divisions

- **Meiosis I** = The reduction division
  - Reduces the number of chromosomes from 46 to 23
- **Meiosis II** = The equational division
  - Produces four cells from the two produced in Meiosis I

Note = Each division contains a prophase, a metaphase, an anaphase and a telophase

Meiosis

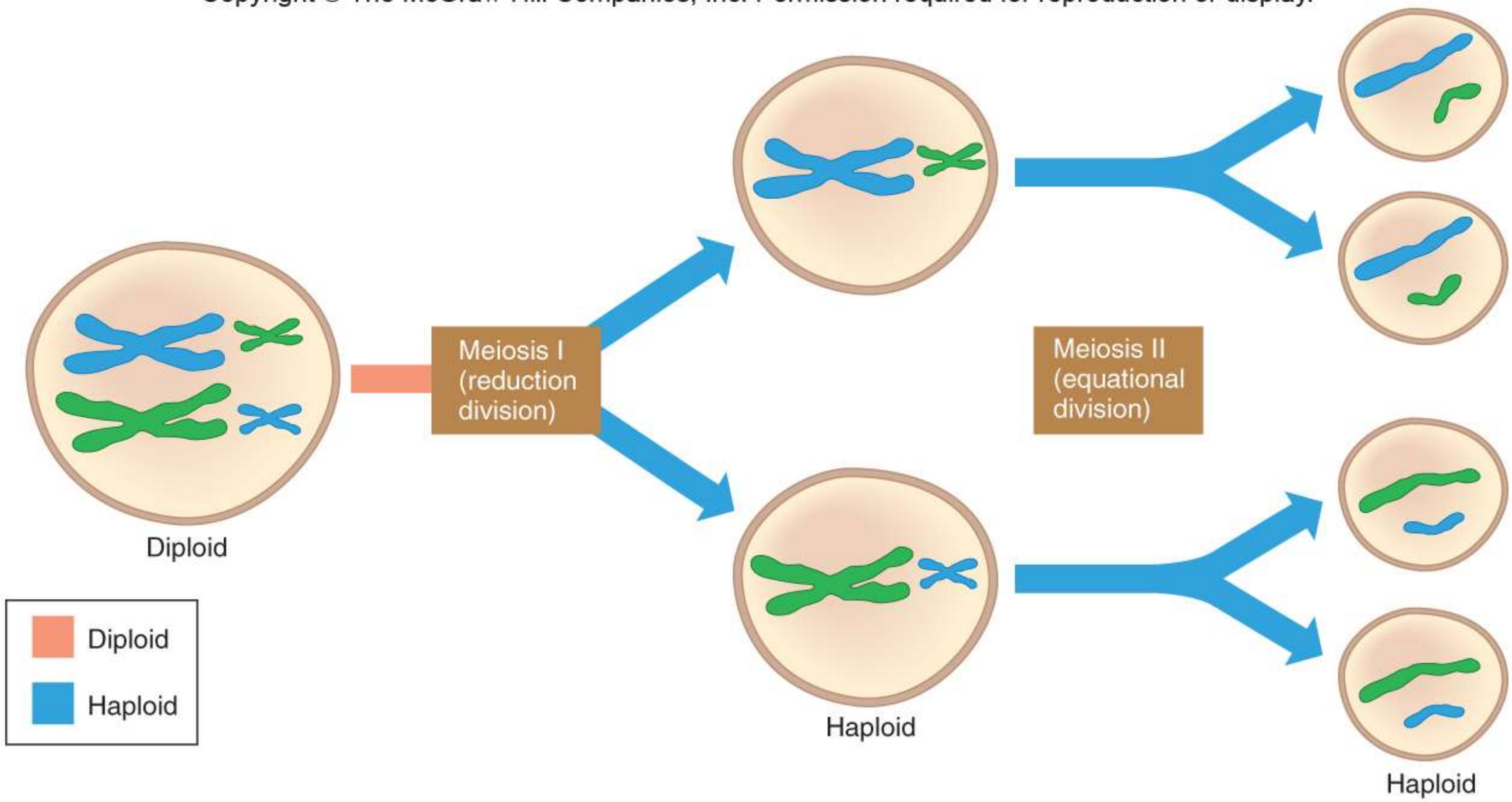
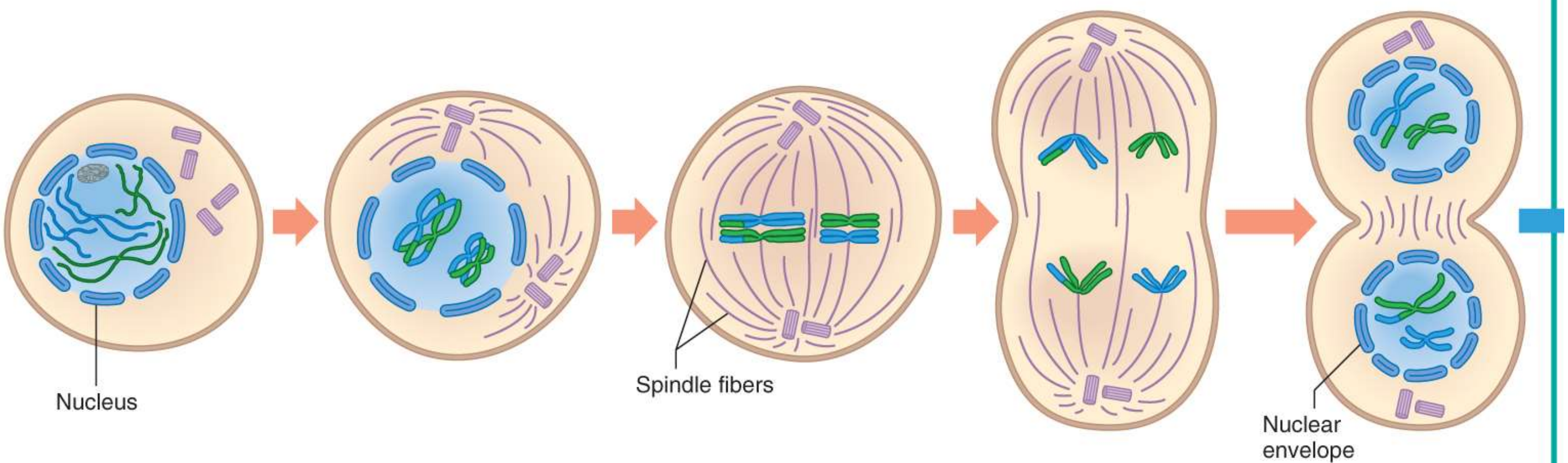


Figure 3.3

Meiosis I

MEIOSIS I

Figure 3.4



Prophase I (early)
Synapsis and crossing over occurs.

Prophase I (late)
Chromosomes condense, become visible. Spindle forms. Nuclear envelope fragments. Spindle fibers attach to each chromosome.

Metaphase I
Paired homologous chromosomes align along equator of cell.

Anaphase I
Homologous chromosomes separate to opposite poles of cell.

Telophase I
Nuclear envelopes partially assemble around chromosomes. Spindle disappears. Cytokinesis divides cell into two.

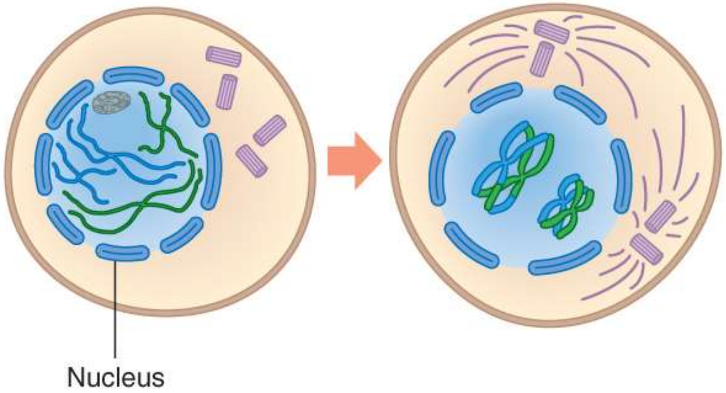
Prophase I

Homologs pair-up and undergo crossing over

Chromosomes condense

Spindle forms

Nuclear envelope breaks down



Prophase I (early)
Synapsis and crossing over occurs.

Prophase I (late)
Chromosomes condense, become visible. Spindle forms. Nuclear envelope fragments. Spindle fibers attach to each chromosome.

Crossing-over

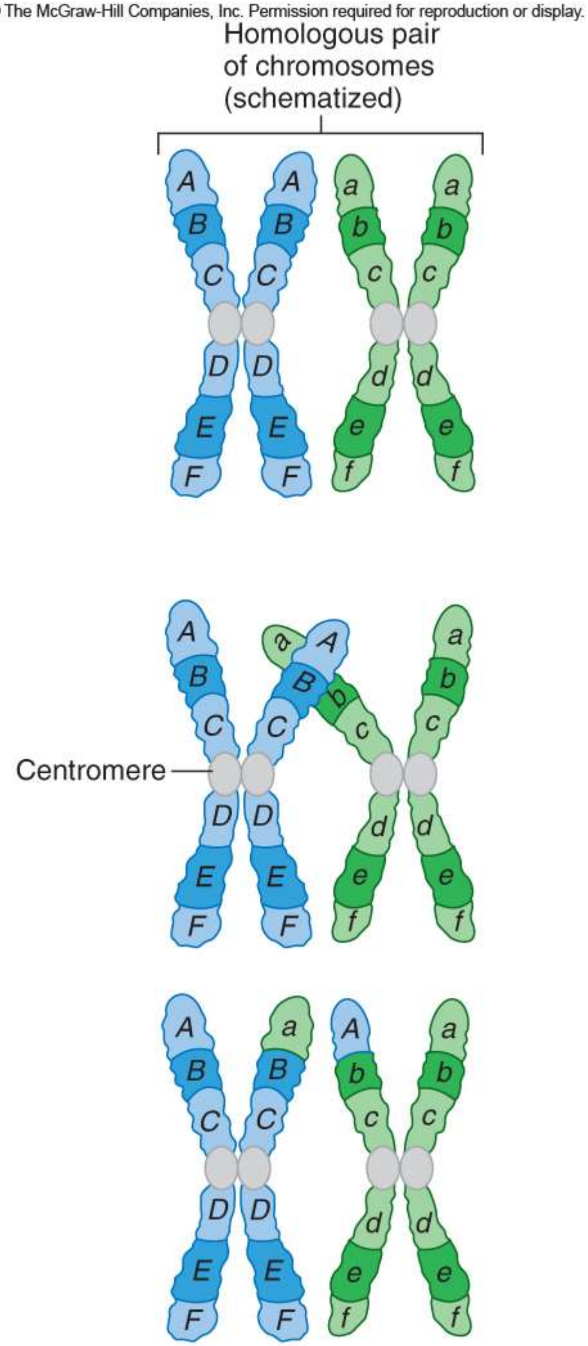
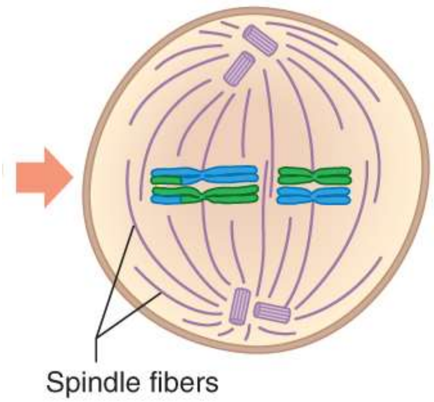


Figure 3.5

Metaphase I

Homologous pairs align along the equator of the cell

The random alignment pattern determines the combination of maternal and paternal chromosomes in the gametes



Metaphase I
Paired homologous chromosomes align along equator of cell.

Independent Assortment

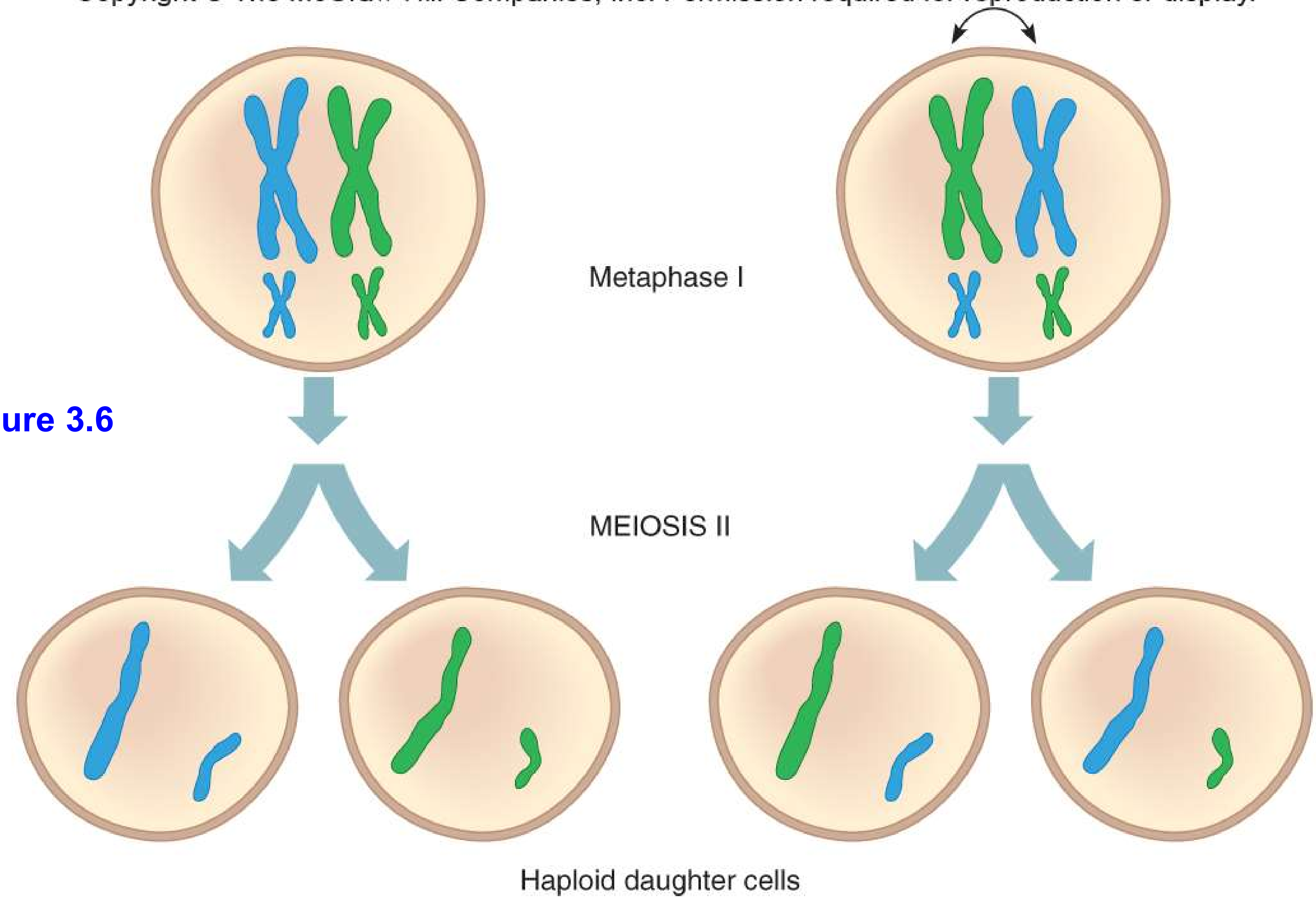
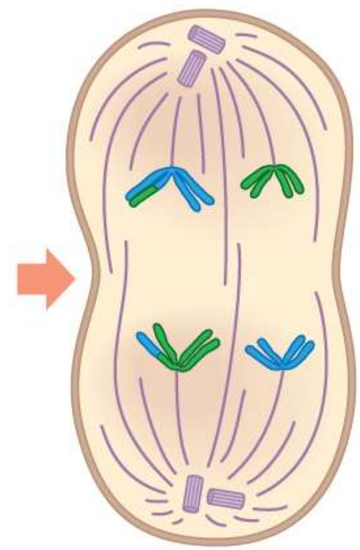


Figure 3.6

Anaphase I

Homologs separate and move to opposite poles of the cell

Sister chromatids remain attached at their centromeres



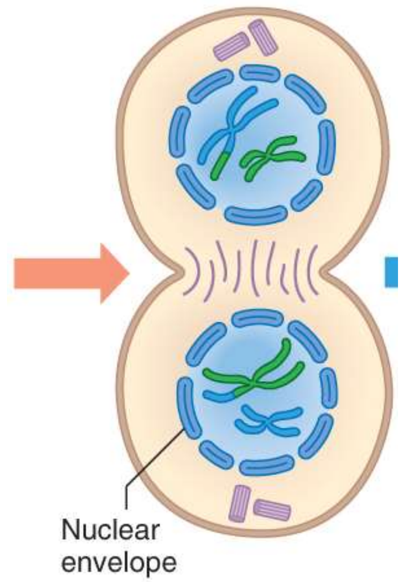
Anaphase I
Homologous chromosomes separate to opposite poles of cell.

Telophase I

Nuclear envelope reforms

Spindle disappears

Cytokinesis divides cell into two



Telophase I
Nuclear envelopes partially assemble around chromosomes. Spindle disappears. Cytokinesis divides cell into two.

Interkinesis

A short interphase between the two meiotic divisions

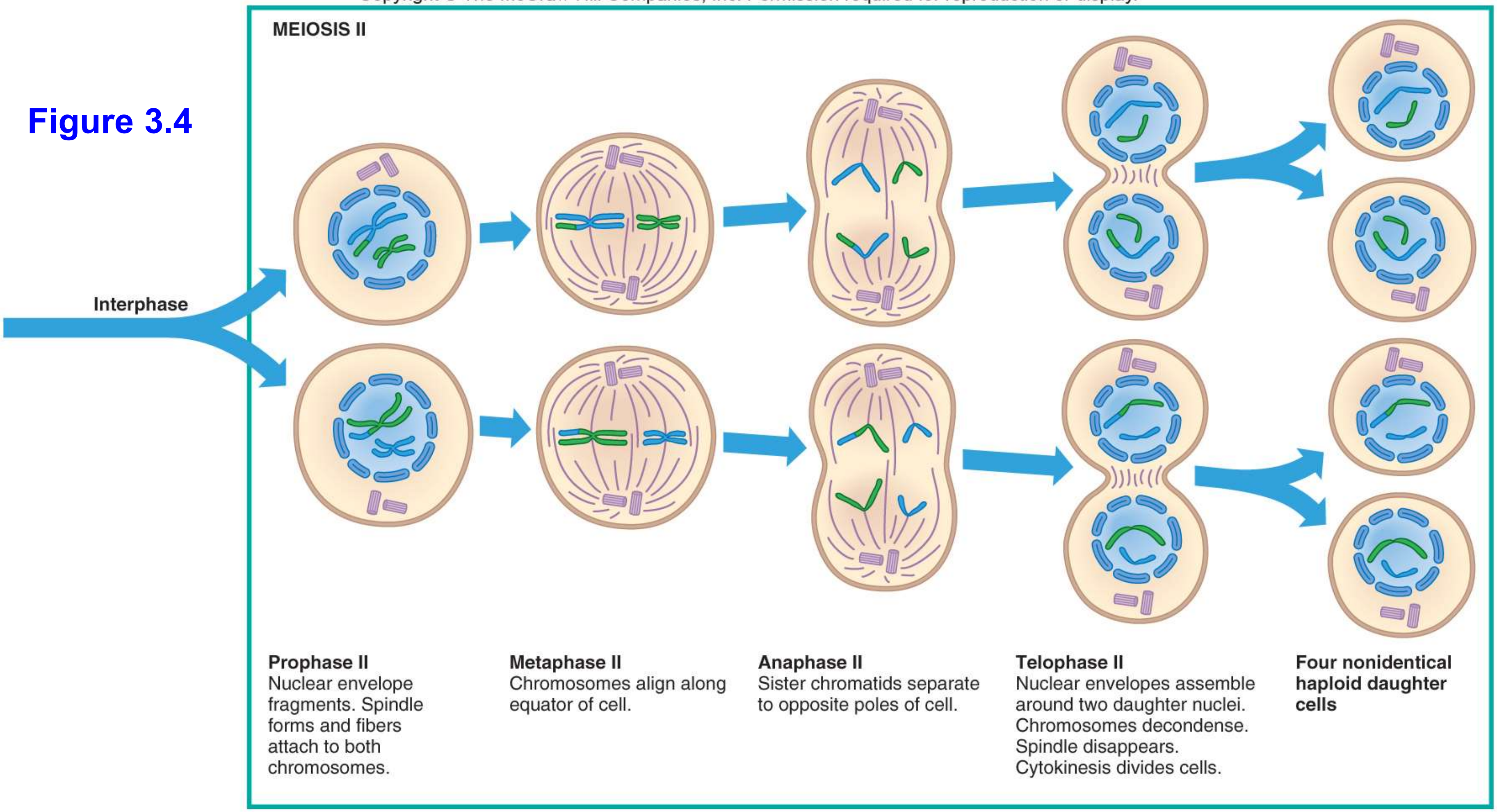
Chromosomes unfold into very thin threads

Proteins are manufactured

However, DNA is NOT replicated a second time

Meiosis II

Figure 3.4



Prophase II

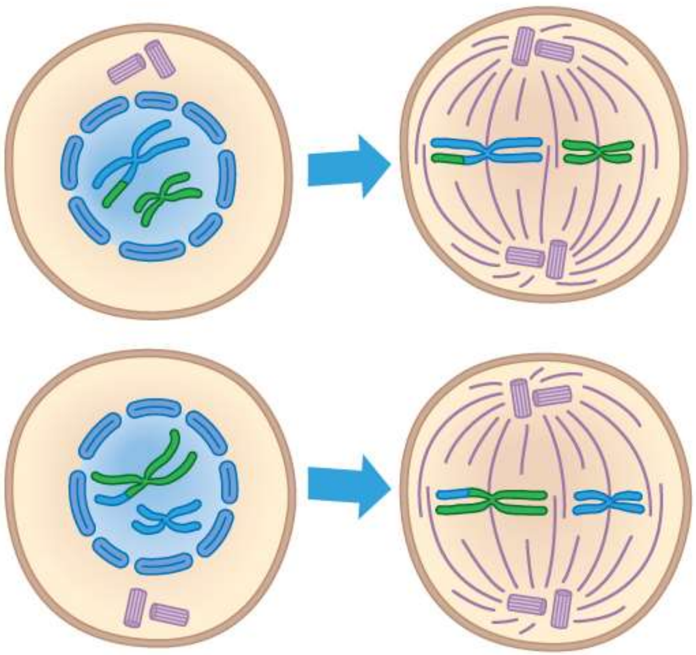
Chromosomes are again condensed and visible

Spindle forms

Nuclear envelope fragments

Metaphase II

Chromosomes align along the equator of the cell



Anaphase II

Centromeres divide

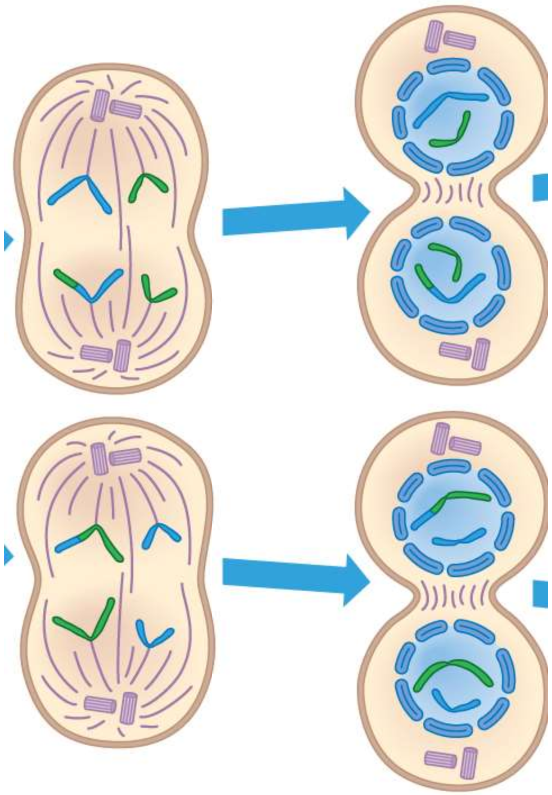
Sister chromatids separate to opposite cell poles

Telophase II

Nuclear envelope reforms

Chromosomes uncoil

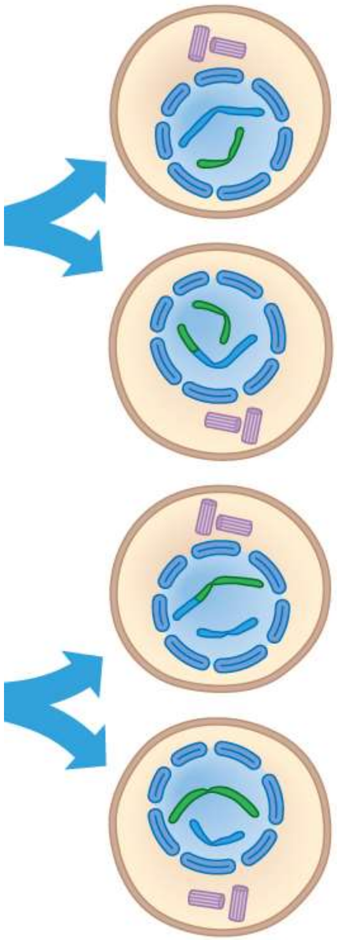
Spindle disappears



Results of Meiosis

Four haploid cells containing a single copy of the genome

Each cell is unique – carries a new assortment of genes and chromosomes



Four nonidentical haploid daughter cells

Comparison of Mitosis and Meiosis

Table 3.1 Comparison of Mitosis and Meiosis

Mitosis	Meiosis
One division	Two divisions
Two daughter cells per cycle	Four daughter cells per cycle
Daughter cells genetically identical	Daughter cells genetically different
Chromosome number of daughter cells same as that of parent cell (2*n*)	Chromosome number of daughter cells half that of parent cell (1*n*)
Occurs in somatic cells	Occurs in germline cells
Occurs throughout life cycle	In humans, completes after sexual maturity
Used for growth, repair, and asexual reproduction	Used for sexual reproduction, producing new gene combinations

Spermatogenesis

A diploid **spermatogonium** divides by mitosis to produce a stem cell and another cell that specializes into a **primary spermatocyte**

In meiosis I, the primary spermatocyte produces two haploid **secondary spermatocytes**

In meiosis II, each secondary spermatocyte produces two haploid **spermatids**

Spermatids then mature into a tad-pole shaped **spermatozoa**

Spermatogenesis

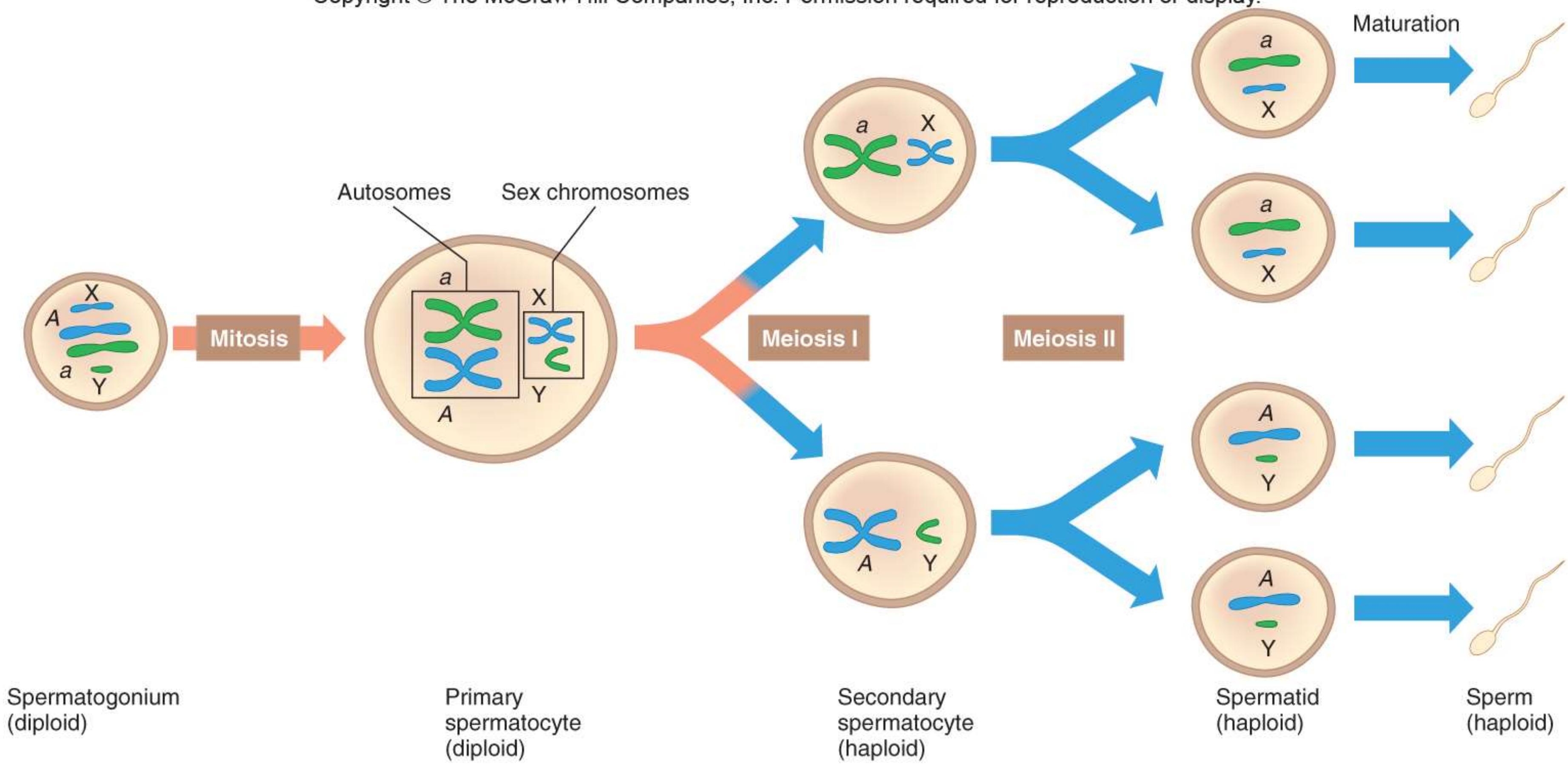


Figure 3.7

Spermatogenesis

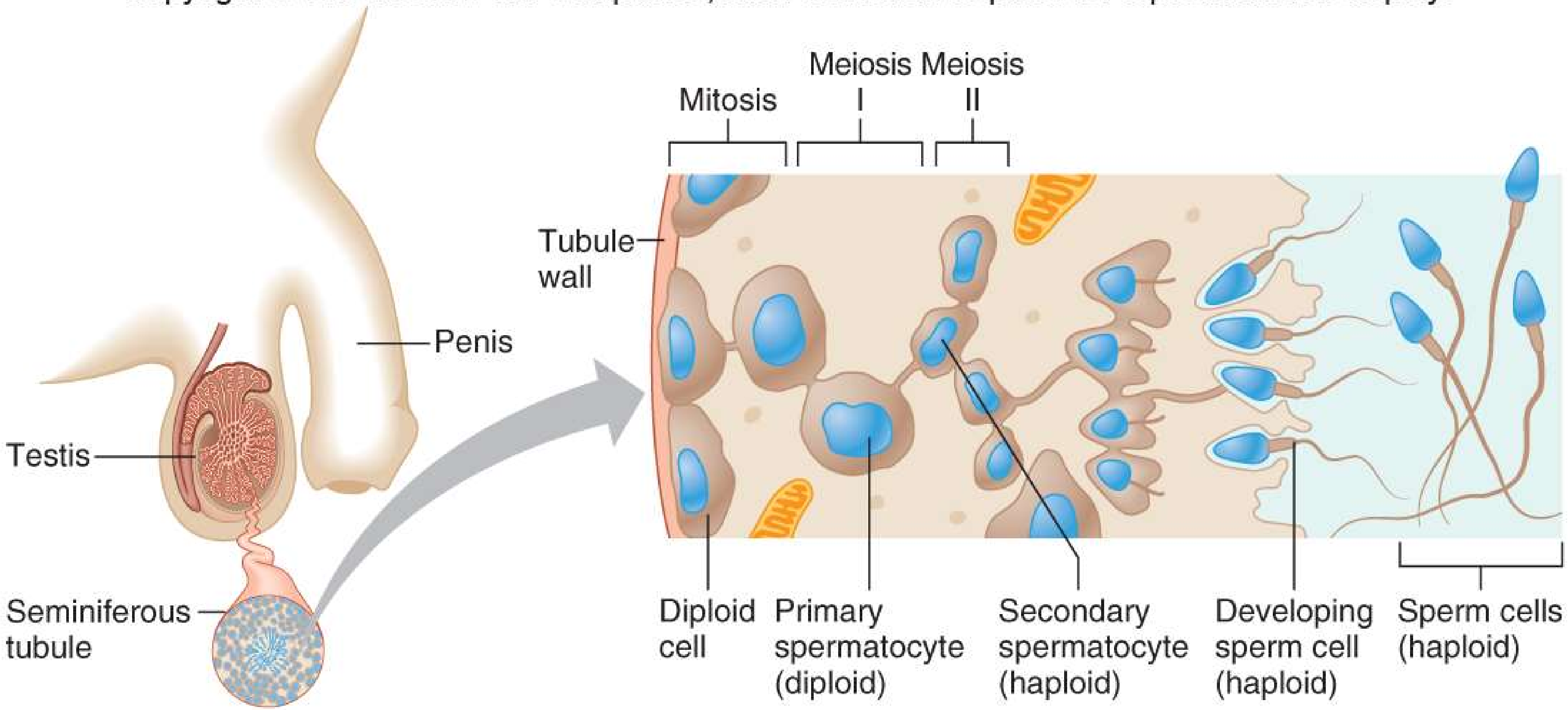
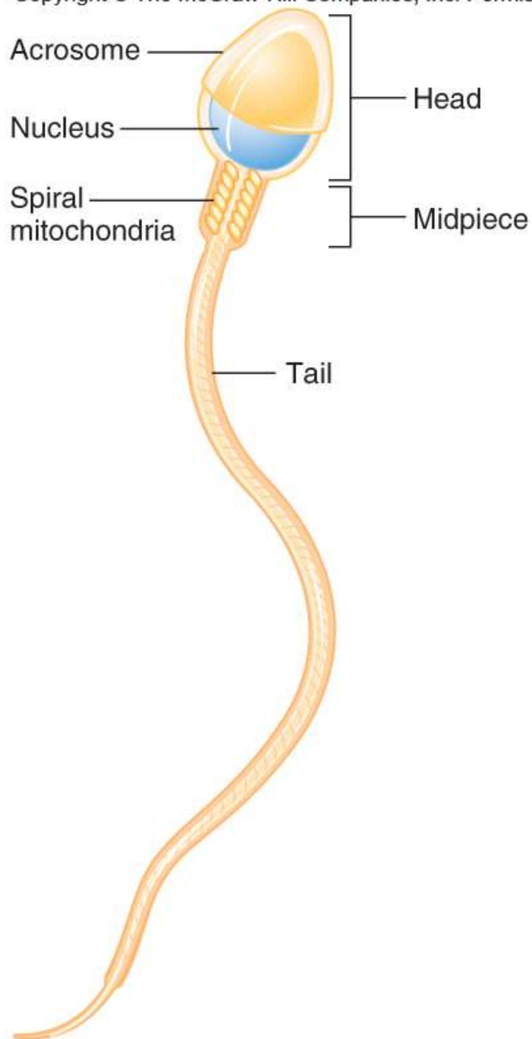


Figure 3.8

Spermatogenesis



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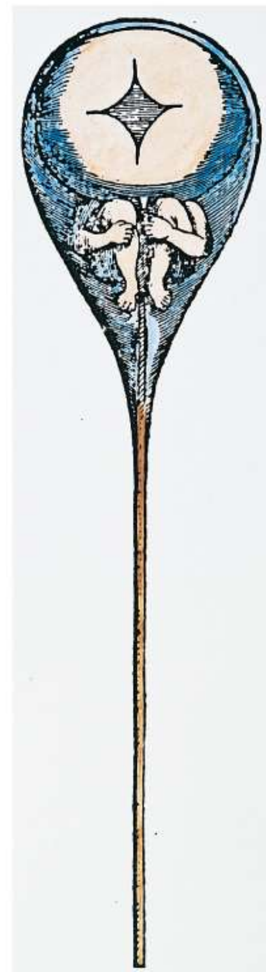

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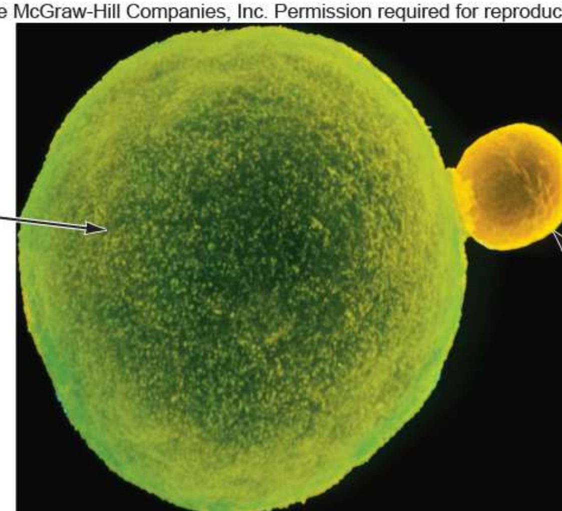
Figure 3.9

Oogenesis

A diploid **oogonium** divides by mitosis to produce a stem cell and another cell that specializes into a **primary oocyte**

In meiosis I, the primary oocyte divides unequally forming a small **polar body** and a large **secondary oocyte**



In meiosis II, the secondary oocyte divides to form another **polar body** and a mature haploid **ovum**

Figure 3.10

Oogenesis

Unlike spermatogenesis, oogenesis is a discontinuous process

A female begins meiosis when she is a fetus

- Oocytes arrest at prophase I until puberty
- After puberty, meiosis I continues in one or several oocytes each month but halts again at metaphase II
- Meiosis is only completed if the ovum is fertilized

Oogenesis

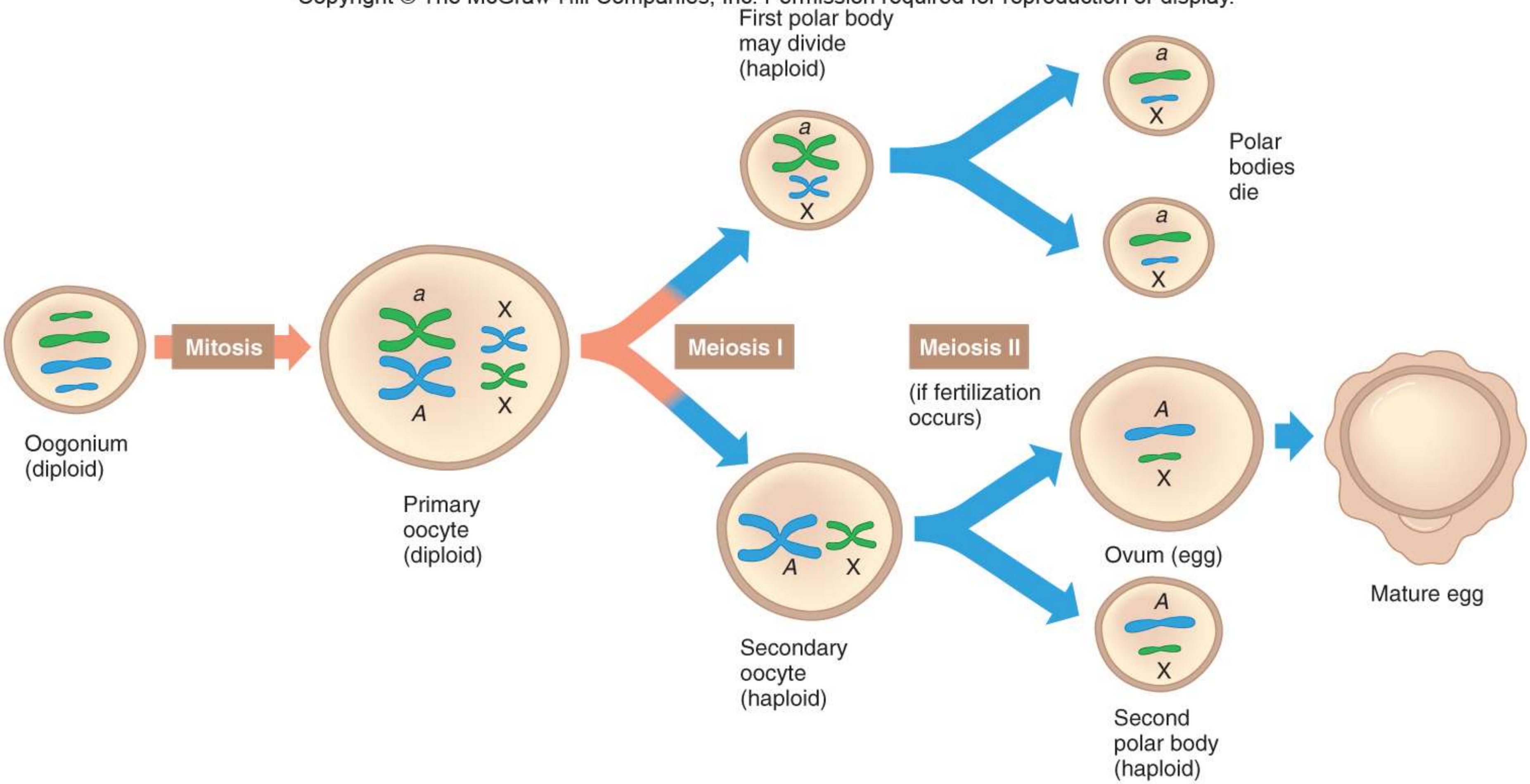


Figure 3.11

Oogenesis

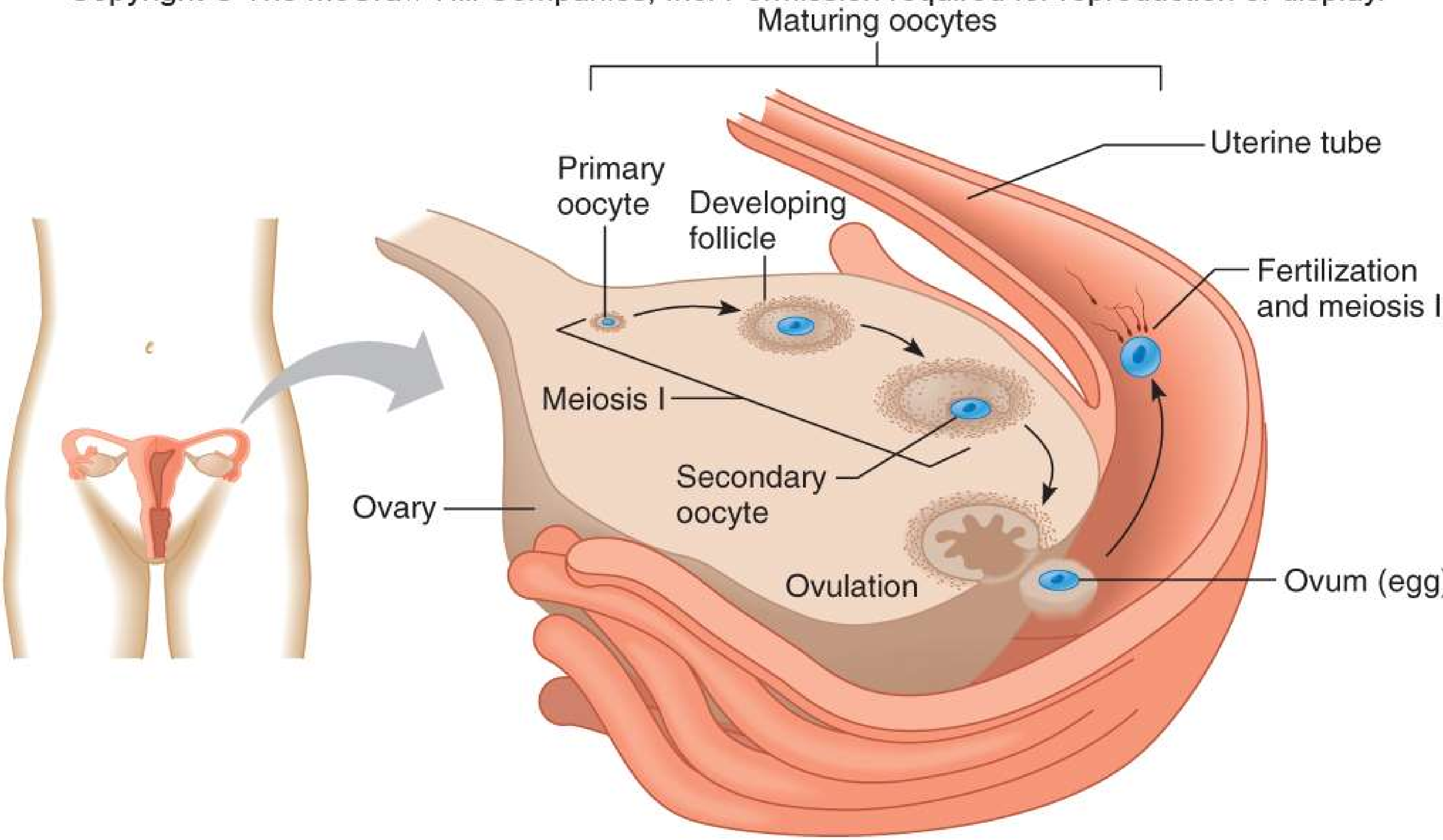


Figure 3.12

Fertilization

Union of sperm and ovum

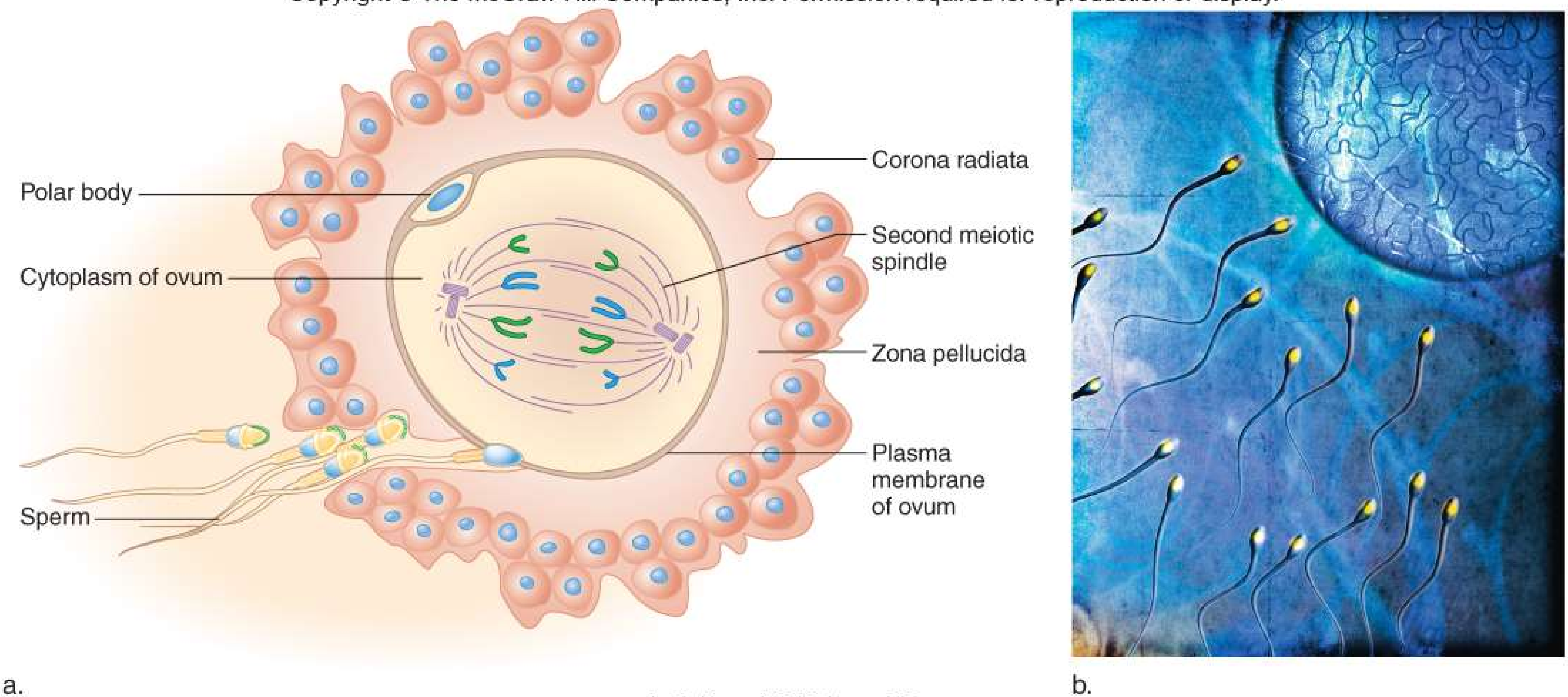
In the female, sperm are capacitated and drawn to the secondary oocyte

Acrosomal enzymes aid sperm penetration

Chemical and electrical changes in the oocyte surface block entry of more sperm

The two sets of chromosomes fuse into one nucleus, forming the **zygote**

Fertilization



b: © Brand X Pictures RF

Figure 3.13

Cleavage

A period of frequent mitotic divisions

- Resulting cells are called **blastomeres**

Developing embryo becomes a solid ball of 16+ cells called a **morula**

The ball of cells hollows out to form a **blastocyst**

Blastocyst

Consists of two main parts

- **Inner cell mass (ICM)**, which develops into the embryo
- **Trophoblast**, which develops into the placenta

Implantation in the uterus occurs around day 7

Certain blastocyst cells secrete human chorionic gonadotropin (hCG)

- A sign of pregnancy

From Ovulation to Implantation

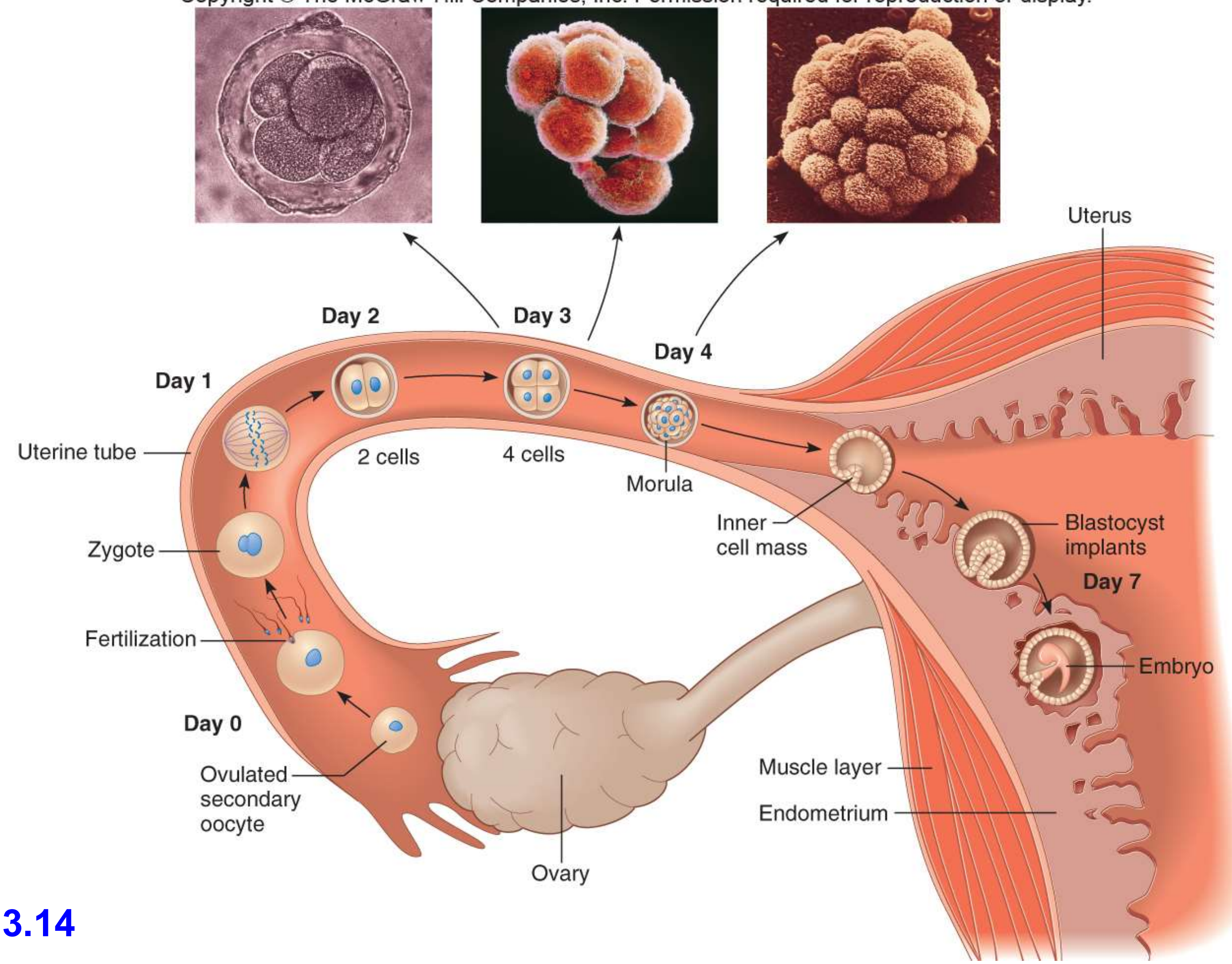


Figure 3.14

Gastrulation

The primary germ layers form in the second week after fertilization

- **Ectoderm** (outermost layer)
- **Mesoderm** (middle layer)
- **Endoderm** (innermost layer)

This three-layered structure is the **gastrula**

Cells in each germ layer begin to form specific organs

Supportive Structures

Structures that support and protect the embryo include:

- Chorionic villi
- Yolk sac
- Allantois
- Umbilical cord
- Amniotic sac

By 10 weeks the placenta is fully formed

The Primordial Embryo

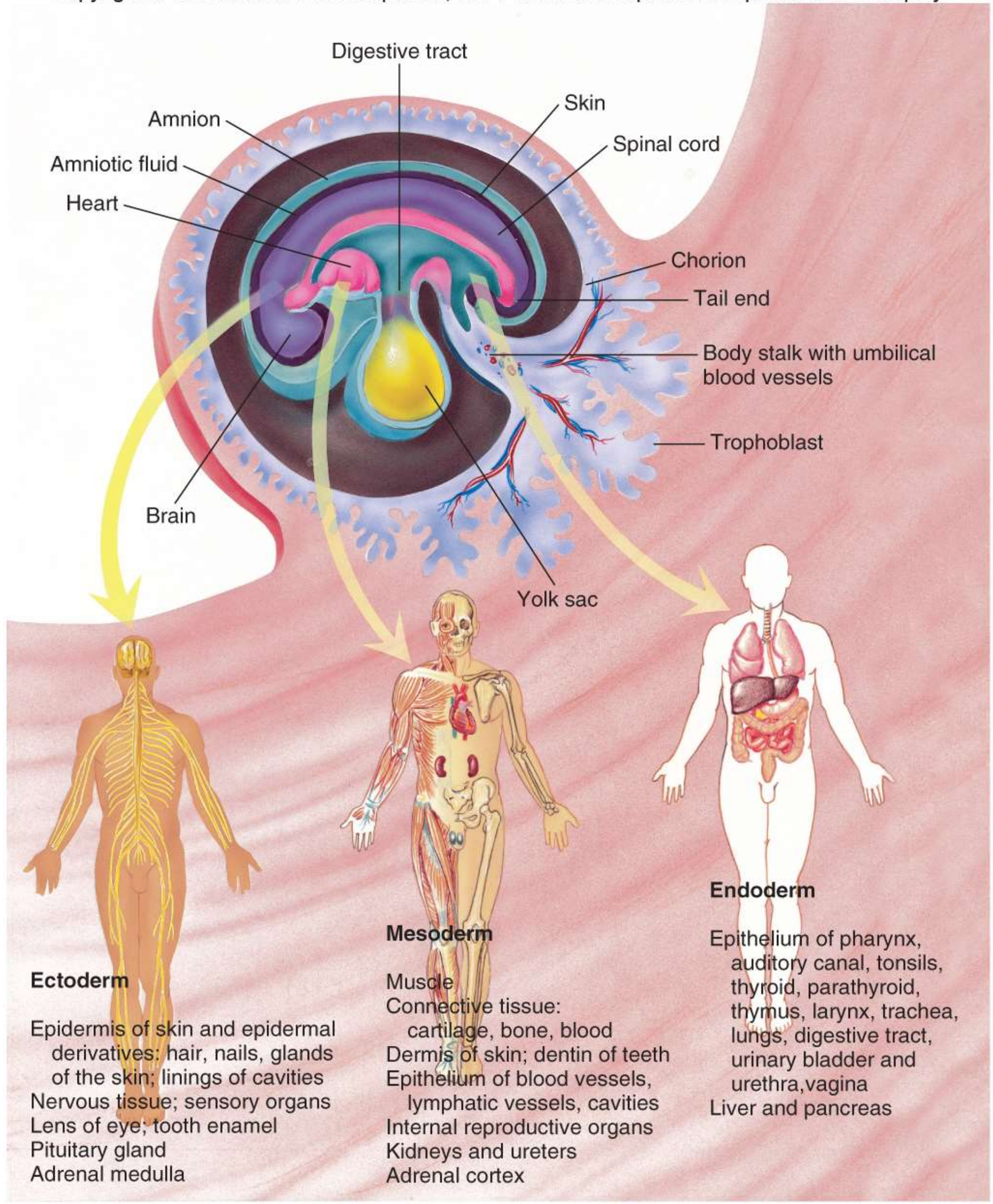


Figure 3.15

Stages of Prenatal Development

Table 3.2 Stages and Events of Early Human Prenatal Development

Stage	Time Period	Principal Events
Fertilized ovum	12–24 hours following ovulation	Oocyte fertilized; zygote has 23 pairs of chromosomes and is genetically distinct
Cleavage	30 hours to third day	Mitosis increases cell number
Morula	Third to fourth day	Solid ball of cells
Blastocyst	Fifth day through second week	Hollowed ball forms trophoblast (outside) and inner cell mass, which implants and flattens to form embryonic disc
Gastrula	End of second week	Primary germ layers form

Multiple Births

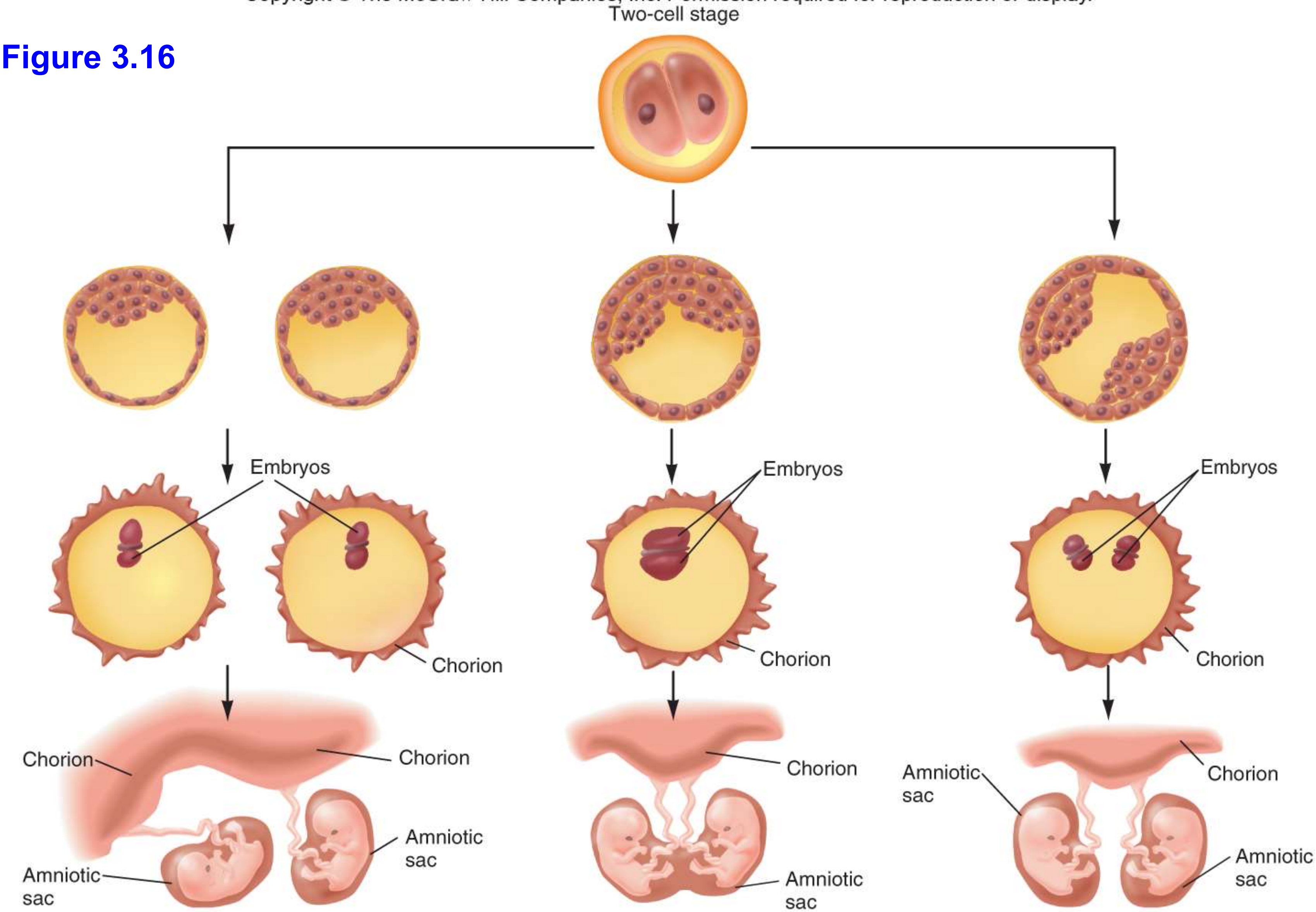
Dizygotic twins (Fraternal)

- Arise from two fertilized ova
- Same genetic relationship as any two siblings

Monozygotic twins (Identical)

- Arise from a single fertilized ovum
- Embryo splits early during development
- Twins may share supportive structures

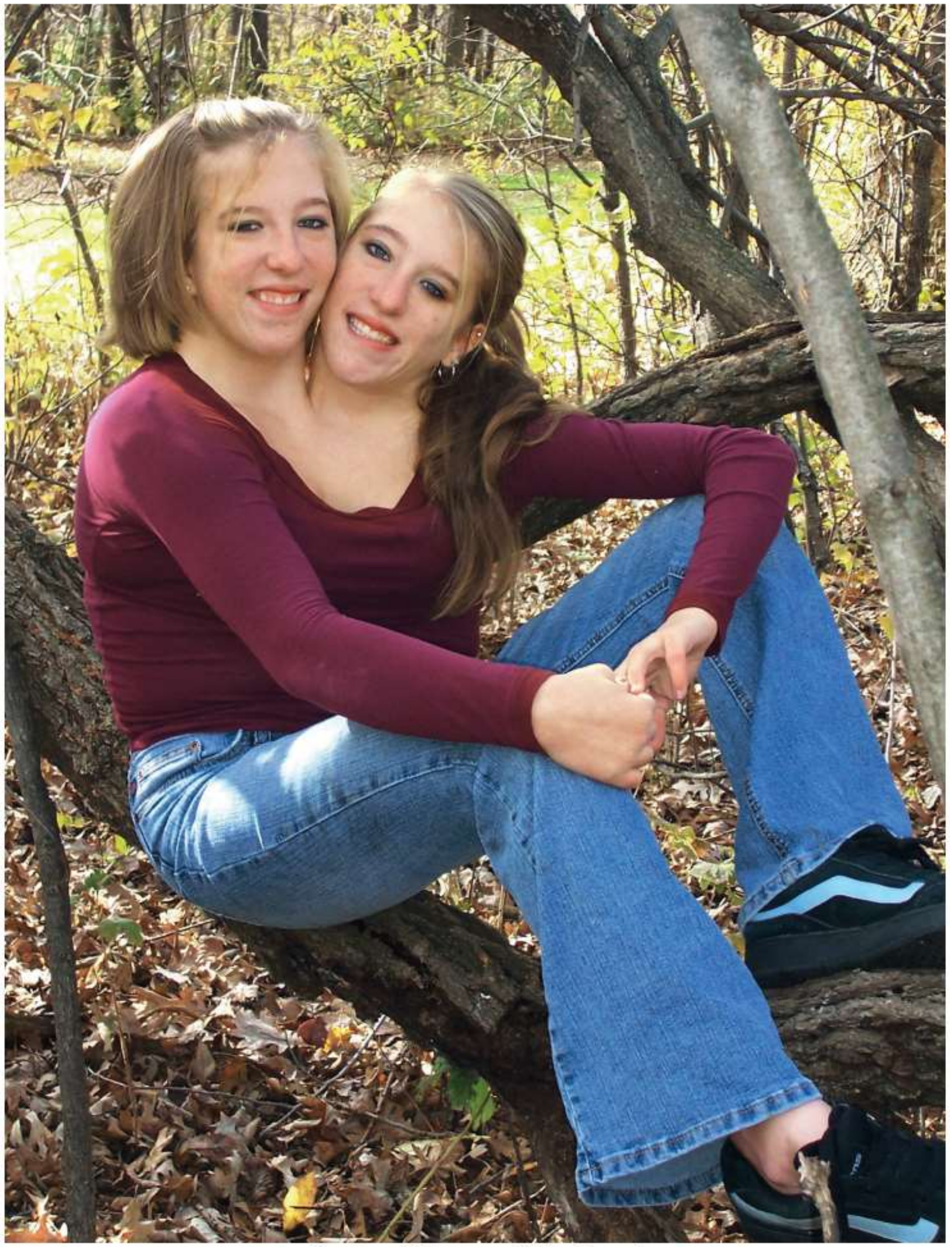
Figure 3.16



a. Identical twins with separate amnions and chorions

b. Identical twins that share an amnion and chorion

c. Identical twins that share a chorion but have separate amnions



Courtesy, Brittany and Abby Hensel

Figure 3.17

The Embryo Develops

Organogenesis is the transformation of the simple three germ layers into distinct organs

During week 3, a band called the primitive streak appears along the back of the embryo

This is followed rapidly by the notochord, neural tube, heart, central nervous system, limbs, digits, and other organ rudiments

By week 8, all the organs that will be present in the newborn have begun to develop

- The prenatal human is now called a **fetus**

The Embryo Develops

a. 28 days 4–6 mm b. 49 days 13–22 mm

a: © Petit Format/Nestle/Photo Researchers;
b: © Petit Format/SPL/Photo Researchers

Figure 3.18

The Fetus Grows

During the fetal period, structures grow, specialize and begin to interact

Bone replaces cartilage in the skeleton

Body growth catches up with the head

Sex organs become more distinct

In the final trimester, the fetus moves and grows rapidly, and fat fills out the skin

The digestive and respiratory systems mature last

Birth Defects

The time when a particular structure is sensitive to damage is called its **critical period**

Birth defects can result from a faulty gene or environmental insult

Most birth defects develop during the embryonic period

- These are more severe than those that arise during the fetal period

Critical Periods of Development

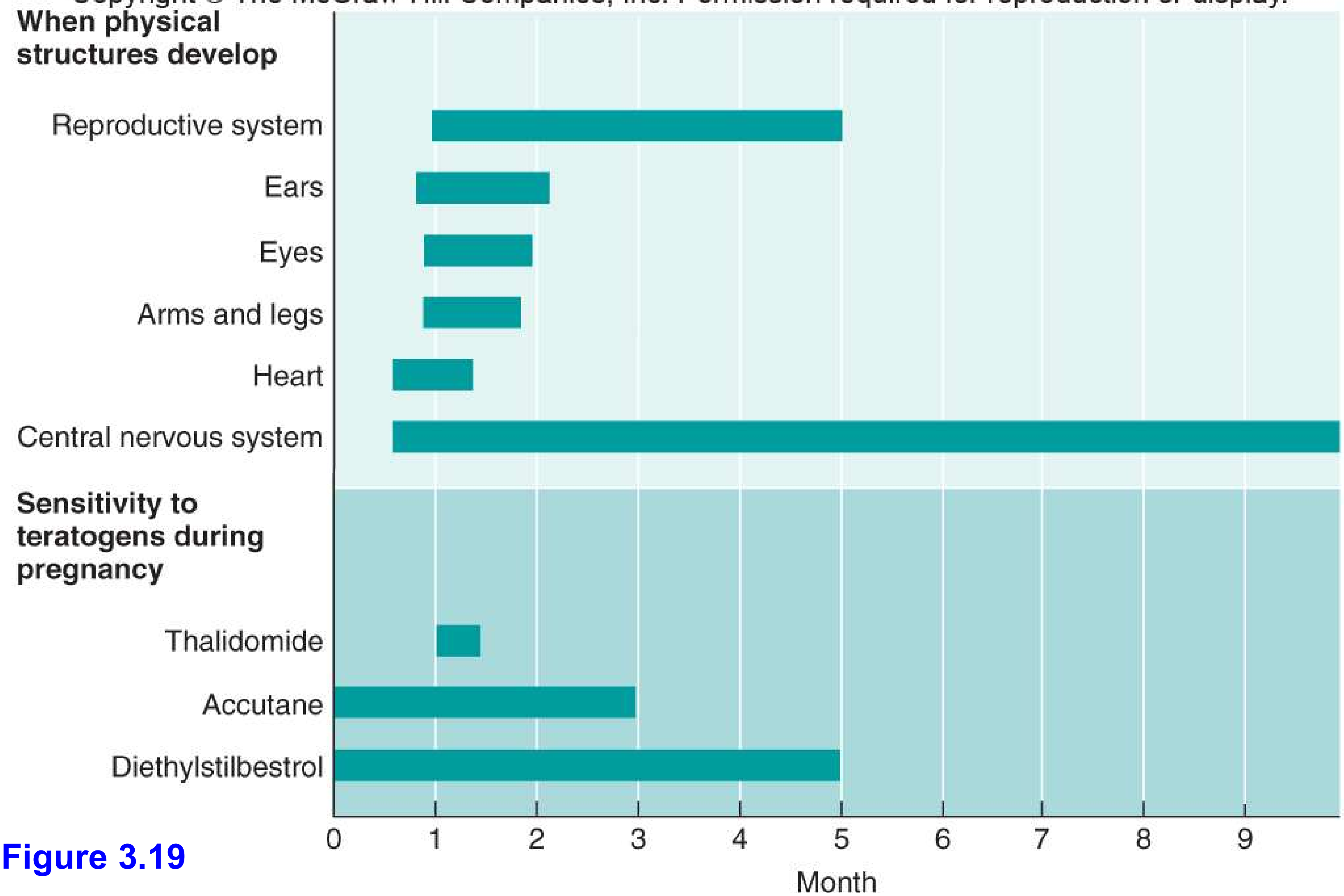


Figure 3.19

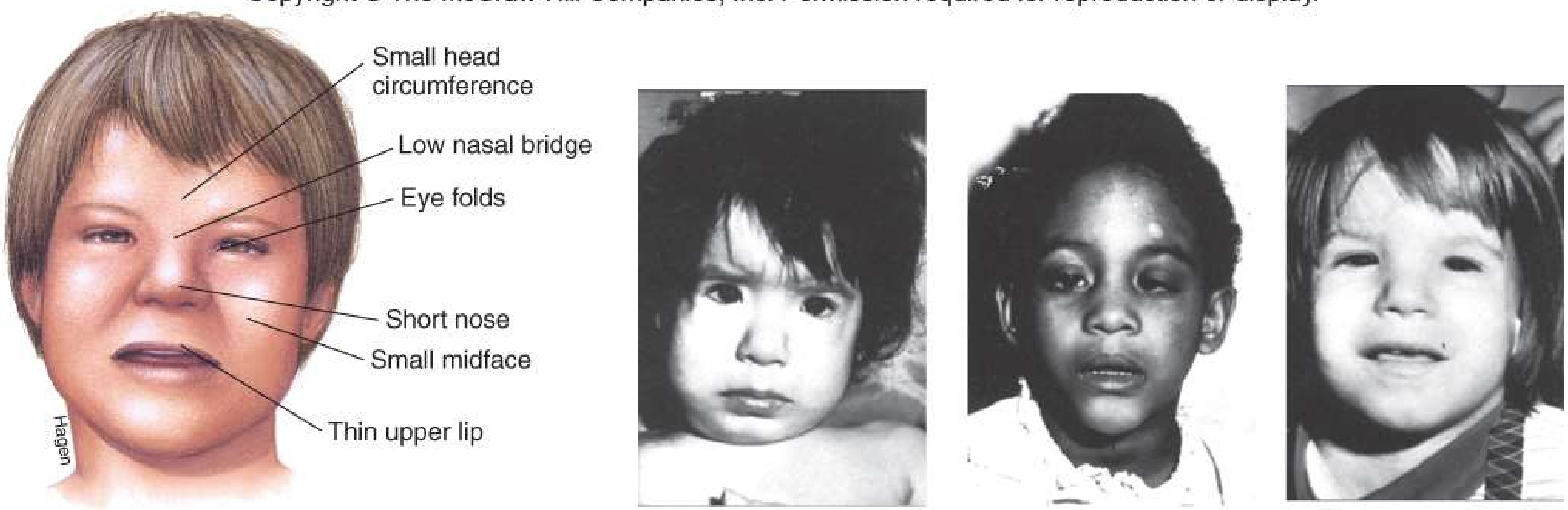
Teratogens

Chemical or other agents that cause birth defects

Examples

- Thalidomide
- Cocaine
- Cigarettes
- Alcohol
- Some nutrients
- Some viruses

Fetal Alcohol Syndrome



a. b. c. d.

From Streissguth, A.P., Landesman-Dwyer, S., Martin, J.C., & Smith, D.W. July 1980. "Teratogenic effects of alcohol in human and laboratory animals." *Science*, 209(18):353-361. @1980 American Association for the Advancement of Science

Figure 3.20

Aging

Genes may impact health throughout life

Single-gene disorders that strike in childhood tend to be recessive

Adult-onset single-gene traits are often dominant

Interaction between genes and environmental factors

- Example: Malnutrition before birth

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Figure 3.21

Aging

Genes control aging both passively and actively

A few single-gene disorders can speed the signs of aging

Segmented progeroid syndromes

- Hutchinson-Guilford syndrome

Photo Courtesy The Progeria Research Foundation

Figure 3.22

Is Longevity Inherited?

Aging reflects genetic activity plus a lifetime of environmental influences

Human chromosome 4 houses longevity genes

-Genome-wide screens of 100-year olds are identifying others

Adoption studies compare the effects of genes vs. the environment on aging