

1

Learning Outcomes

1. Describe the structures of the male and female reproductive systems.
2. Explain why meiosis is necessary to reproduce.
3. Summarize the events of meiosis.
4. List the steps in sperm and oocyte formation.
5. Describe early prenatal development.
6. Explain how the embryo differs from the fetus.
7. Define *critical period*.
8. List some teratogens.
9. Describe common diseases that begin in adulthood.
10. Explain how rapid aging syndromes occur.

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Human Development

Genes orchestrate our physiology after conception through adulthood.

First cell forms when a **sperm** from a male and an **oocyte** from a female join.

- Sperm and oocytes are **gametes**.

Each reproductive system has:

- Paired structures, called gonads
- Tubular structures that transport these cells
- Hormones and secretions that control reproduction

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The Male Reproductive System 1

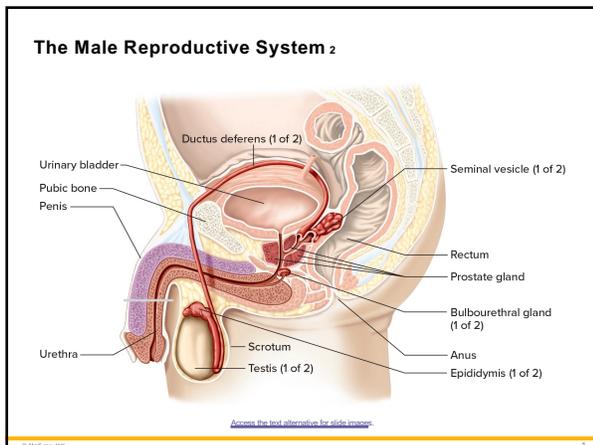
Sperm cells are made in the seminiferous tubules of the **testes**.

Sperms mature, stored in the epididymis.

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

Sperm from each ductus deferens exit through the urethra and out the penis.

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The Female Reproductive System 1

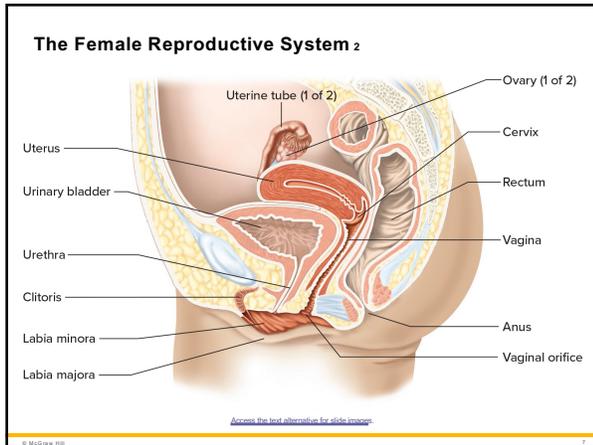
Oocytes mature in the **ovaries**.

Each month, an ovary releases an oocyte into one of two **uterine tubes**.

- 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 maturation and the preparation of the uterus to nurture a fertilized ovum.

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Meiosis 1

Gametes form from special cells called germ line cells

Meiosis is a cell division that halves the chromosome number

- **Homologous pairs** have the same genes in the same order but carry different alleles, or variants, of the same gene

Gametes are **haploid** and somatic cells are **diploid** for each chromosome

Absence of meiosis could lead to genetically overloaded cells

Mixes up trait combinations

Provides genetic diversity which can enable a population to survive an environmental challenge

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Meiosis 2

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:

- As in mitosis, meiosis occurs after an interphase period when DNA is replicated (doubled)
- Each division contains a prophase, a metaphase, an anaphase, and a telophase

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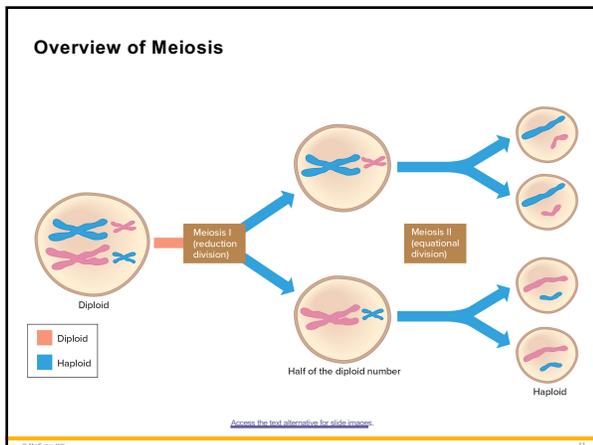
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Mitosis vs Meiosis

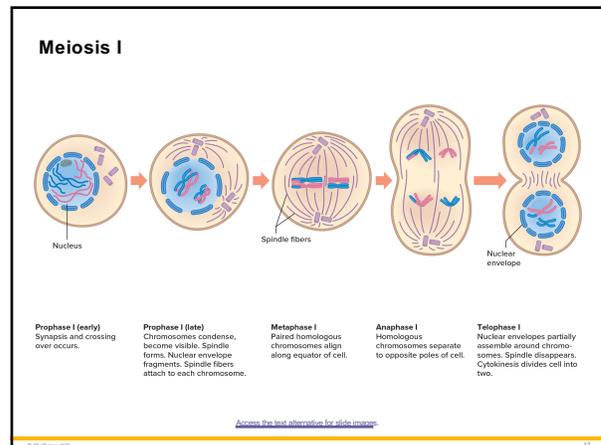
Mitosis	Meiosis
One division	Two divisions
Two daughter cells per cycle	Four daughter cells per cycle
Daughter cells genetically identical	Daughter cells genetically different
Chromosome number of daughter cells same as that of parent cell (2n)	Chromosome number of daughter cells half that of parent cell (1n)
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

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Prophase I₁

A spindle forms.

Homologs pair-up and undergo **crossing over**.

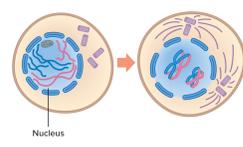
Chromosomes condense.

Synapsed chromosomes separate but remain attached at a few points.

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Prophase I₂



Prophase I (early)

- Synapsis and crossing over occurs.

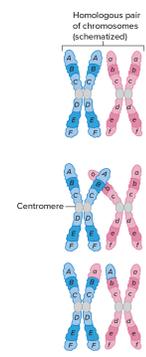
Prophase I (late)

- Chromosomes condense, become visible. Spindle forms. Nuclear envelope fragments. Spindle fibers attach to each chromosome.

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Crossing Over



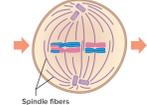
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Metaphase I

Homologous pairs align along the equator of the cell.

Random alignment of chromosomes causes **independent assortment** of the genes that they carry.



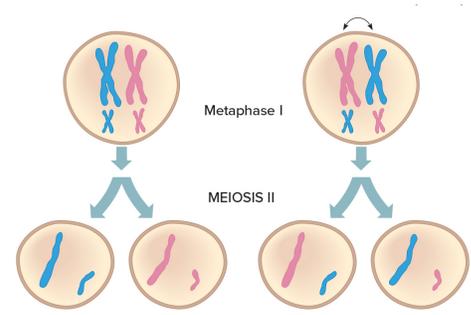
Metaphase I

- Paired homologous chromosomes align along equator of cell.

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Independent Assortment



Haploid daughter cells

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Anaphase I and Telophase I₁

Homologs separate in anaphase I

- Unlike in mitosis, the centromeres of each replicated chromosome in meiosis I remain together.

Homologs move to opposite poles by telophase I

Note:

- During a second interphase, chromosomes unfold into thin threads.
- Proteins are manufactured, but DNA is not replicated a second time.

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Anaphase I and Telophase I

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.

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Meiosis II

Prophase II Metaphase II Anaphase II Telophase II Four nonidentical haploid daughter cells

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Prophase II and Metaphase II

Chromosomes are again condensed and visible.
Chromosomes align along the equator of the cell.

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Anaphase II and Telophase II

Centromeres divide
Newly formed, unreplicated chromosomes, move to opposite poles
Nuclear envelope reforms
Separate into individual cells

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Results of Meiosis 1

Four nonidentical haploid daughter cells

- Each carries a new assortment of genes and chromosomes that hold one copy of the genome

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Results of Meiosis 2

Meiosis generates astounding genetic variety.

- A person can produce 2^{23} (8,388,608) possible combinations of chromosomes

Thus, fertilization of gametes can generate more than 70 trillion ($8,388,608 \times 8,388,608$) genetically unique individuals!

Crossing over contributes almost limitless genetic diversity.

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Gametes Mature

Meiosis happens in both sexes, but different distributions of cell components create the distinctions between sperm and oocytes.

The gametes of the maturing male and female proceed through similar stages as they form, but with vastly different timetables.

- A male begins manufacturing sperm at puberty and continues throughout life, whereas a female begins meiosis when she is a fetus.
- Meiosis in the female completes only if a sperm fertilizes an oocyte.

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Spermatogenesis 1

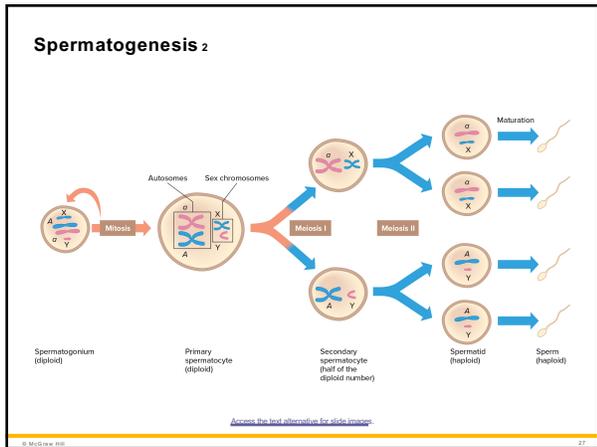
A diploid **spermatogonium** divides by mitosis to produce a stem cell and another cell that specializes into a mature sperm.

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

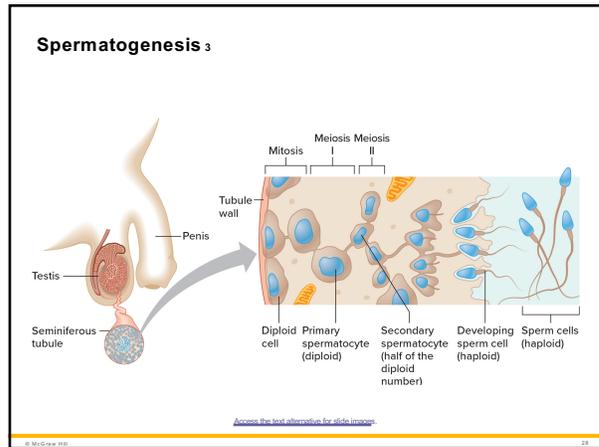
In meiosis II, each secondary spermatocyte produces two equal-sized **spermatids**.

Spermatids then mature into tadpole-shaped **spermatozoa**.

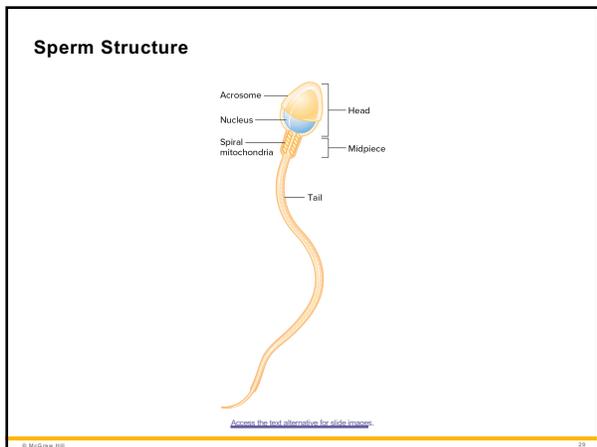
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Oogenesis 1

Begins with a diploid **oogonium**

In meiosis I, 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 **ovum**

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Oogenesis 2

Unlike spermatogenesis, oogenesis is a discontinuous process.

Oocytes arrest at prophase I until puberty.

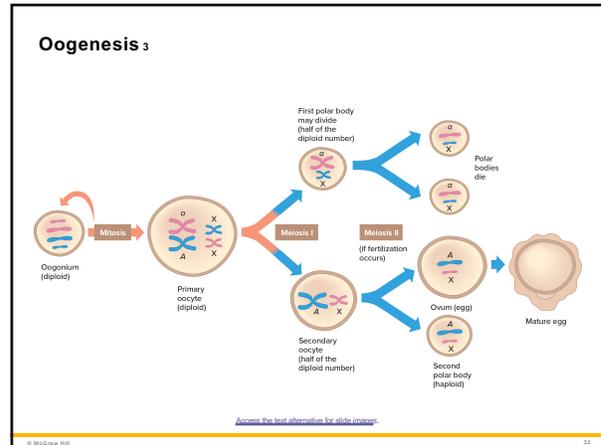
After puberty, meiosis I continues in one or several oocytes each month but halts again at metaphase II.

Meiosis is only completed if the ovum is fertilized.

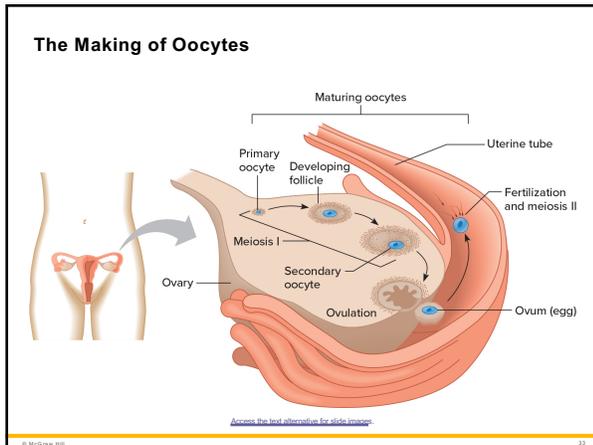
A female ovulates about 400 oocytes between puberty and menopause.

- Most oocytes degrade, because fertilization is so rare.

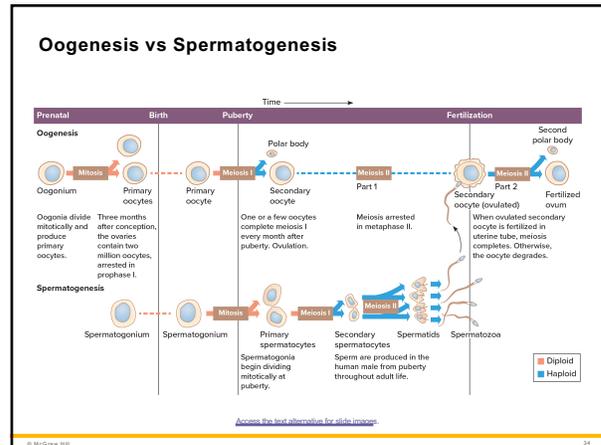
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Meiosis and Mutations

“Paternal age effect” conditions arise from stem cells in testes that divide every 16 days, from puberty on.

- Thus offering many opportunities for DNA replication error that generate a dominant mutation.

Mutations in the fibroblast growth factor receptor (FGFR) arise more frequently in testes as a man ages.

- This skews meiosis, producing problems in skeletal growth.

Genetic errors in oocytes from older women are typically extra or absent chromosomes.

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Paternal Age Effect

Disease	Phenotype
Achondroplasia	Short-limbed dwarfism (see figure 5.1a)
Crouzan syndrome	Premature fusion of skull bones in infancy, causing wide-spaced and bulging eyes, beaked nose, short upper lip, small upper jaw, and jutting lower jaw
Hutchinson-Gilford progeria syndrome	Thin hair, weak bones, tough and wrinkled skin, stiff joints and blood vessel linings
Multiple endocrine neoplasia 2	Cancers of thyroid, parathyroid, and adrenal glands
Pfeiffer syndrome	Premature fusion of skull bones in infancy, short and fused fingers and toes
Thanatophoric dysplasia	Severe short-limbed dwarfism

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Prenatal Development

A prenatal human is considered an **embryo** for the first 8 weeks, when rudiments of all body parts form.

- The embryonic period begins when the fertilized ovum divides by mitosis.
- During the first week, the embryo is in a "preimplantation" stage because it has not yet settled into the uterine lining.

Prenatal development after the eighth week is the period when structures grow and specialize.

- From the start of the 9th week until birth, the prenatal human organism is a **fetus**.

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Fertilization 1

Union of sperm and oocyte

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

Acrosomal enzymes aid sperm penetration

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

Two genetic packages meet and merge, forming a **zygote**

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Fertilization 2

Labels: Polar body, Corona radiata, Second meiotic spindle, Cytoplasm of ovum, Zona pellucida, Plasma membrane of ovum, Sperm.

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Cleavage

A day after fertilization, the zygote divides by mitosis, beginning a period of frequent cell division called **cleavage**

- Resulting early cells are called **blastomeres**

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

The ball of cells hollows out, and its center fills with fluid, creating a **blastocyst**

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Blastocyst

Some of the blastocyst cells form a clump on the inside lining called the **inner cell mass**.

- Develops into the embryo.

Outermost blastocyst cells form the **trophoblast**

- Secrete the hormone human chorionic gonadotropin (hCG) that prevents menstruation.
- A sign of pregnancy

Implantation in the uterus occurs around day 7.

- Takes about a week

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From Ovation to Implantation

Labels: Uterine tube, Uterus, Muscle layer, Endometrium, Ovary, Day 0 (Ovulated secondary oocyte), Day 1 (Fertilization), Day 2 (2 cells), Day 3 (4 cells), Day 4 (Morula), Day 7 (Blastocyst implantation), Embryo, Inner cell mass.

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Embryo Formation

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 layer begin to form specific organs controlled by genes called homeotic

Epigenetic effects oversee differentiation as cells in each germ layer develop into organs.

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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 from the chorionic villi

- It continues to secrete hormones that maintain pregnancy and sends nutrients to the fetus

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The Primordial Embryo

The diagram shows a cross-section of a developing embryo with various organs labeled. Arrows point from these organs to three human figures below, each representing a different germ layer: Ectoderm, Mesoderm, and Endoderm. The Ectoderm list includes skin, hair, nails, and the nervous system. The Mesoderm list includes muscle, bone, blood, and the circulatory system. The Endoderm list includes the digestive tract, lungs, and internal organs.

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Stages of Early Prenatal Development

Stage	Time Period	Principal Events
Fertilized ovum	12 to 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 fluid-filled 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

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Multiple Births

Twins & other multiples arise early in development

Dizygotic twins (DZ; Fraternal)

- Two sperm fertilize two oocytes
- Same genetic relationship as any two siblings

Monozygotic twins (MZ; Identical)

- Arise from a single fertilized ovum
- Three types of MZ twins can form, depending upon when the fertilized ovum or very early embryo splits
- Exposed to slightly different uterine environments

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Types of Identical Twins

The diagram shows a 'Two-cell stage' at the top. Three arrows lead to different developmental paths for identical twins. Path (I) shows two separate embryos, each with its own amniotic sac and chorion. Path (II) shows two embryos sharing a single amniotic sac but having separate chorions. Path (III) shows two embryos sharing both a single amniotic sac and a single chorion.

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Conjoined Twins



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The Embryo Develops 1

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

Followed by the connective tissue progenitor cells, notochord, **neural tube**, heart, central nervous system, arms, legs and other organ rudiments

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

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The Embryo Develops 2



28 days (a) 4-6 mm 49 days (b) 13-22 mm

© McGraw Hill (a) Paul Fournelle/Science Source; (b) Paul Fournelle/Science Source

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The Fetus Grows 1

During the fetal period, body proportions approach those of a newborn

Bone replaces softer cartilage

Nerve and muscle functions become coordinated

Anatomical differences between the sexes appear at week 6

- After the *SRY* gene is expressed in males.

By week 12, sucks thumb, kicks, makes fists and faces, and has the beginnings of teeth

By the fourth month, the fetus has hair, eyebrows, lashes, nipples, and nails

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The Fetus Grows 2

Vocal cords will be formed by 18 weeks.

- But the fetus makes no sound because it doesn't breathe air.

By the end of the second trimester, the woman feels distinct kicks and jabs and may detect fetal hiccup.

In the final trimester, fetal brain cells link into networks as organs elaborate and grow, and fat fills out the skin.

The digestive and respiratory systems mature last.

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16-Week Fetus



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Birth Defects

The time when genetic abnormalities, toxic substances, or viruses can alter a specific structure is called the **critical period**

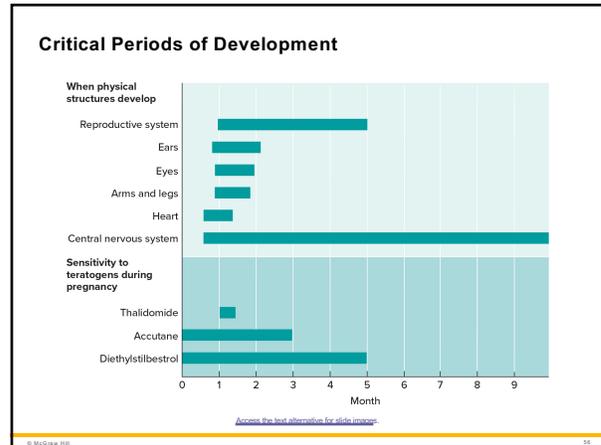
About two-thirds of birth defects develop during the embryonic period

- More severe than those that arise during the fetal period

Some birth defects are caused by a mutation that acts at a specific point in prenatal development.

- Phocomelia is a suppression of limb development

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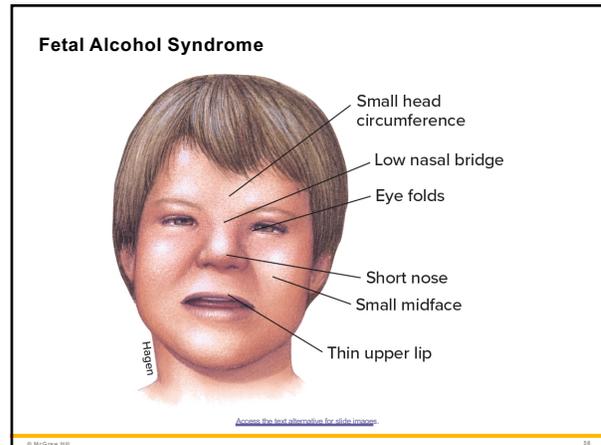
Teratogens

Are chemical or other agents that cause birth defects

Examples:

- Cocaine
- Cigarettes
- Thalidomide
- Alcohol
 - Fetal alcohol syndrome
- Nutrients
- Vitamins
- Viral infections
 - Zika virus; HIV; German measles (rubella)

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Maturation and Aging

“Aging” means moving through the life cycle

Age 30 seems to be a turning point for decline.

- Some researchers estimate that, after this age, the human body becomes functionally less efficient by about 0.8 percent each year.

Many diseases that begin in adulthood, or are associated with aging, have genetic components.

- These diseases tend to be complex

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Adult-Onset Inherited Disorders

Genes may impact health throughout life.

Environmental factors can affect how certain genes can create risks that appear later.

- A powerful environmental influence is malnutrition.

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

Dominantly inherited conditions affect health in early to middle adulthood.

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Syndromes That Resemble Accelerated Aging

Genes control aging both passively (as structures break down) and actively (by initiating new activities).

Progeroid syndromes are single-gene disorders that speed aging-associated changes.

- The most severe progeroid syndromes are the progerias, which shorten life expectancy.

Most accelerated aging conditions are caused by the inability of cells to adequately repair DNA

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Premature Aging Syndromes

Disease	Incidence	Average Life Expectancy
Ataxia telangiectasia	1/60,000	19–25
Cockayne Syndrome	1/100,000	20
Hutchinson-Gilford progeria syndrome	<1/1,000,000	13
Rothmund- Thomson syndrome	<1/100,000	Normal
Trichothiodystrophy	1/1,000,000	?
Werner syndrome	<1/100,000	50

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Hutchinson-Gilford Progeria Syndrome 1

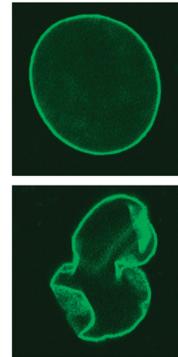
Is the most severe rapid aging condition.

Normal cells growing in culture divide about 50 times before dying.

- Cells from Hutchinson-Gilford progeria syndrome patients die in culture after only 10 to 30 divisions.
- Caused by a mutation in the *LMNA* (lamin) gene
- A sticky form of lamin A, called progerin, triggers rapid programmed cell death (apoptosis)

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Hutchinson-Gilford Progeria Syndrome 2



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Is Longevity Inherited? 1

Aging reflects genetic activity and a lifetime of environmental influences

Families and genetically isolated populations with many aged members have:

- Gene variants
- Shared environmental influences

Genome comparisons among centenarians to others are revealing genes that influence longevity

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Is Longevity Inherited? 2

Centenarians have inherited 2 types of gene variants— those that directly protect them and wild type alleles of genes that, when mutant, cause disease.

Genes can affect longevity control via:

- immune system functioning;
- insulin secretion and glucose metabolism;
- response to stress;
- the cell cycle;
- DNA repair;
- lipid (including cholesterol) metabolism;
- nutrient metabolism; and
- production of antioxidant enzymes.

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Longevity Genes

Gene	Protects Against	Population Studied
Apolipoprotein C3 (APOC-3)	Hypertension, diabetes	Ashkenazi Jews
	Cardiovascular disease	Amish
Bitter taste receptor (TAW2R16)	Poisoning, digestive problems	Calabria, Italy
Cholesteryl ester transfer protein (CETP)	Cardiovascular disease	Ashkenazi Jews
Forkhead box O3 (FOXO3)	Cancer, cardiovascular disease	Japanese-Americans
Growth hormone receptor (GHR)	Diabetes, cancer	Ecuador, Israel
Uncoupling proteins (UCP 2, 3, 4)	Oxidative damage, poor energy use	Calabria, Italy

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