

Principles of Anatomy and Physiology
14th Edition
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WILEY

CHAPTER 29
Development and Inheritance

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Embryonic Period

- The **embryonic period** extends from fertilization through the eighth week of development.
- **Fertilization**—merging of genetic information from **sperm** and **secondary oocyte**.
- Sperm swim from the vagina to the cervix using their tails.
- Sperm pass through the uterus and uterine tubes mainly due to **contraction of the walls of these structures**.

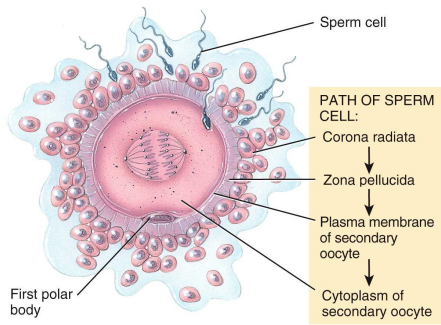
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Embryonic Period

To fertilize an egg, sperm must penetrate the **corona radiata** (granulosa cells) and the **zona pellucida** (glycoprotein layer outside of the oocyte's plasma membrane).

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Embryonic Period



(a) Sperm cell penetrating secondary oocyte

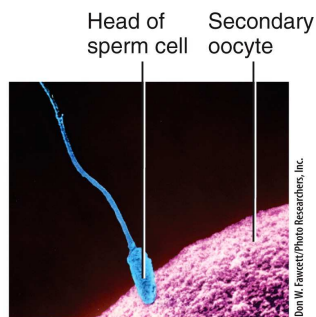
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Embryonic Period

- The enzymes of the sperm's **acrosome**, along with tail movement, allow the sperm to penetrate the **corona radiata**.
- **Glycoprotein ZP3** in the zona pellucida is a receptor for the sperm.
- Membrane proteins in the sperm head bind to ZP3 and **acrosomal enzymes** are released to digest a path in the zona pellucida.

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Embryonic Period



(b) Sperm cell in contact with secondary oocyte

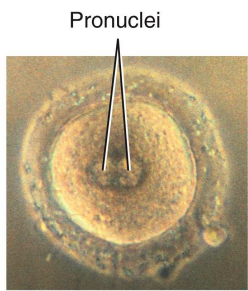
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Embryonic Period

- The haploid nucleus in the head of the sperm becomes the **male pronucleus**.
- The haploid nucleus of the fertilized ovum becomes the **female pronucleus**.
- When the two merge (**syngamy**), the **diploid zygote** is formed.

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Embryonic Period



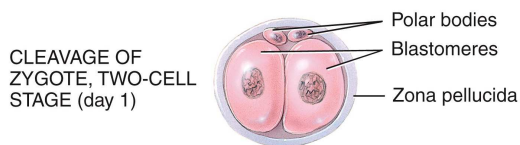
LM 250x

(c) Male and female pronuclei

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Embryonic Period

After fertilization (at about 24 hours), the zygote begins mitotic division called **cleavage**. The first division takes about 6 hours. Successive divisions take less time.

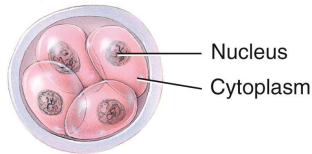


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Embryonic Period

By the second day after fertilization, a **second cleavage** is completed yielding **4 cells**.

CLEAVAGE OF ZYGOTE, FOUR-CELL STAGE (day 2)

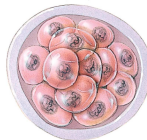


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Embryonic Period

- By the end of the third day there are 16 cells. Each division yields smaller and smaller cells (**blastomeres**).
- By the fourth day the cluster of cells **resembles a mulberry** and is called a **morula**. It is still surrounded by the **zona pellucida** and is still the size of the zygote.

MORULA (day 4)



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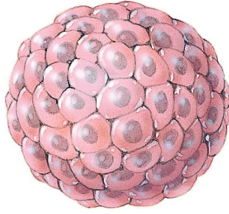
Embryonic Period

- On day 4 or 5, the morula enters the uterine cavity and is nourished by **uterine milk**, a glycogen-rich secretion from endometrial glands in addition to stored nutrients from the cytoplasm.
- At the 32-cell stage, the fluid now inside the morula, rearranges the blastomeres into a large, fluid filled **blastocyst cavity (blastocoel)**. The mass is now called a **blastocyst** (still the same size as the original zygote).

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Embryonic Period

BLASTOCYST,
EXTERNAL VIEW
(day 5)



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Embryonic Period

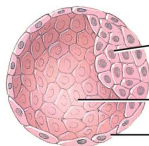
As the blastocyst formed, two different cell populations arose:

- The **embryoblast (inner cell mass)** will develop into the **embryo**.
- The **trophoblast (outer cell mass)** will develop into the outer **chorionic sac** surrounding the fetus, and the **fetal portion of the placenta**.

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Embryonic Period

BLASTOCYST
(sectioned),
INTERNAL VIEW
(day 5)

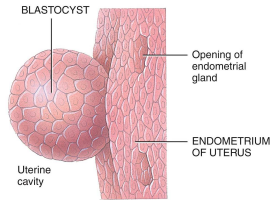


Embryoblast
(inner cell mass)
Blastocyst cavity
Trophoblast

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Embryonic Period

The blastocyst remains free in the uterine cavity for about 2 days and then **implants** by attaching to the **endometrium** at around 6 days after fertilization.



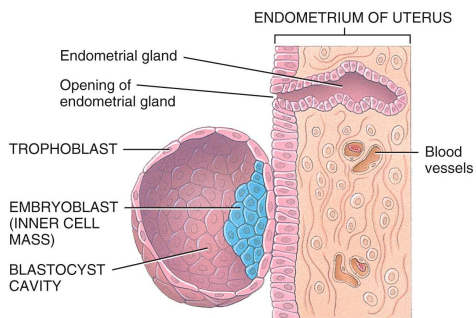
(a) External view of blastocyst, about 6 days after fertilization
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Embryonic Period

- Implantation usually occurs in either the posterior portion of the **fundus** or the **body of the uterus**.
- The **inner cell mass** orients toward the **endometrium**.

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Embryonic Period



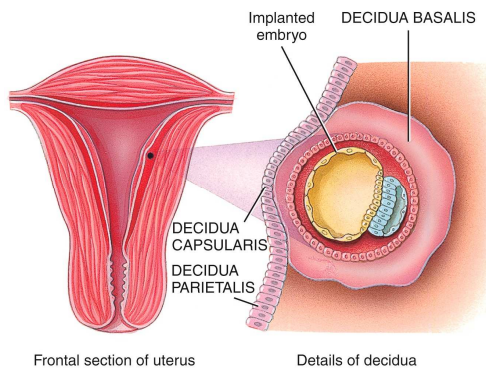
(b) Frontal section through endometrium of uterus and blastocyst, about 6 days after fertilization
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Embryonic Period

- After implantation, the endometrium is called the **decidua**. It separates from the endometrium after the fetus is delivered.
- The decidua has different regions named based on their positions relative to the site of the implanted blastocyst.

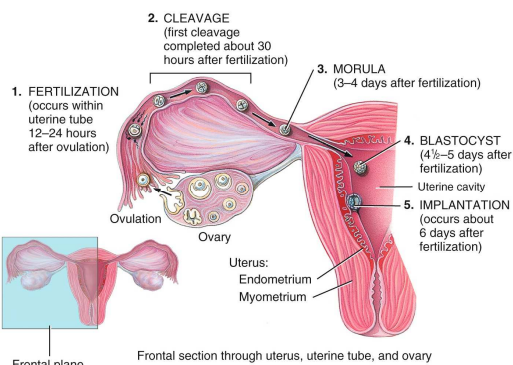
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Embryonic Period



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Embryonic Period



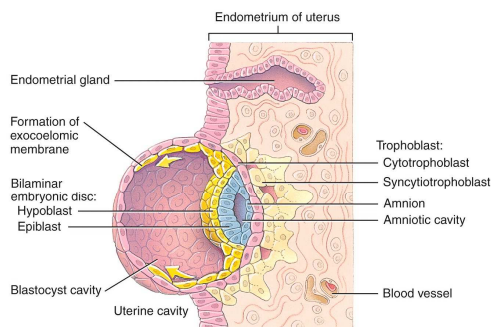
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Embryonic Period

- About **8 days after implantation**, the trophoblast develops into the **syncytiotrophoblast** and **cytotrophoblast**.
- At around 8 days, the **embryoblast** also develops into two layers: the **hypoblast (primitive endoderm)** and **epiblast (primitive ectoderm)**.
- Cells of these structures form a flat disc called the **bilaminar embryonic disc**.
- The **amniotic cavity** forms from the epiblast.

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Embryonic Period



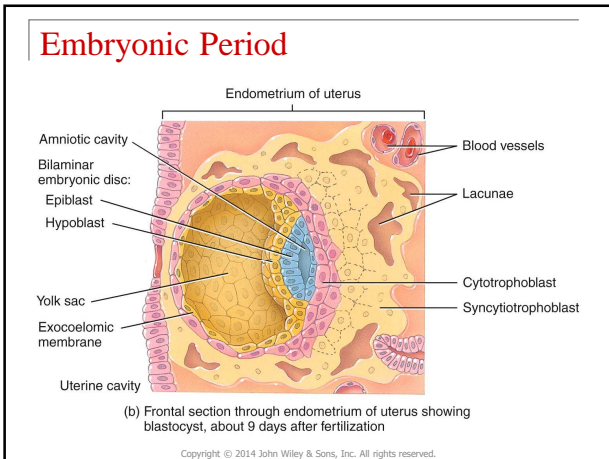
(a) Frontal section through endometrium of uterus showing blastocyst, about 8 days after fertilization

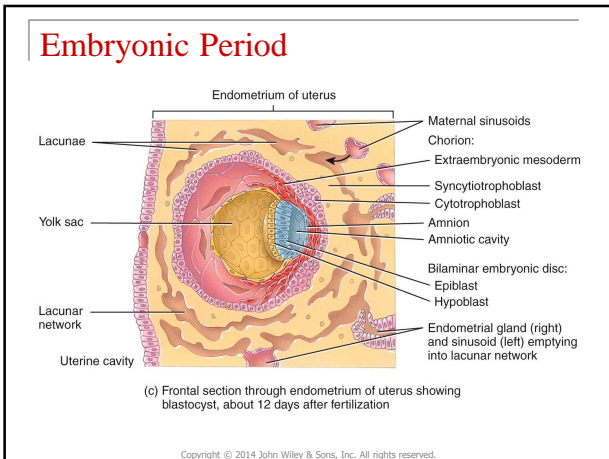
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Embryonic Period

- The **amnion** forms from the roof of the amniotic cavity.
- Eventually, it surrounds the entire embryo and fills with **amniotic fluid**.
- Also on the 8th day, the **exocoelomic membrane** forms that, together with the hypoblast forms the **yolk sac**.
- On the 9th day, small spaces called **lacunae** form.
- By the 12th day, they fuse to form **lacunar networks**.

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Embryonic Period

- About the 12th day after fertilization, the **extraembryonic mesoderm** develops.
- The cells form a connective tissue layer around the **amnion** and **yolk sac**.
- Large cavities develop that fuse and form the **extraembryonic coelom**.

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Embryonic Period

The extraembryonic mesoderm together with the trophoblast forms the **chorion** which surrounds the embryo and, later, the fetus. The chorion

- **Blocks antibody production** by the mother
- Promotes production of **T lymphocytes** to suppress the immune response in the uterus
- Produces **human chorionic gonadotropin (hCG)**

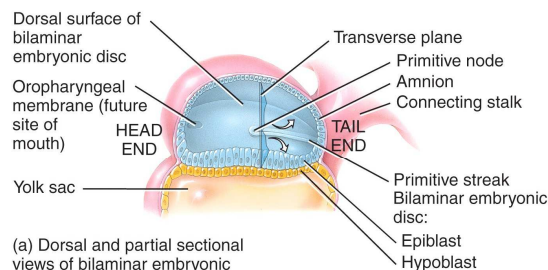
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Embryonic Period

- The first major event of the 3rd week of development is **gastrulation**.
- The two-layered embryonic disc transforms into a **trilaminar (three-layered) embryonic disc (ectoderm, mesoderm, endoderm)**
- Gastrulation is associated with the **rearrangement and migration of cells from the epiblast**.
- The first step in gastrulation is formation of the **primitive streak**.

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Embryonic Period



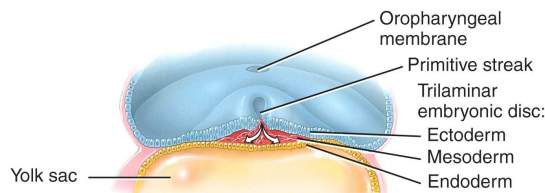
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Embryonic Period

- The primitive streak establishes the **head and tail ends of the embryo**.
- Next, cells of the epiblast move inward below the primitive streak and undergo **invagination**.
- Following this, the **three germ layers form**.

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Embryonic Period



(b) Transverse section of trilaminar embryonic disc, about 16 days after fertilization

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Embryonic Period

TABLE 29.1
Structures Produced by the Three Primary Germ Layers

ENDODERM	MESODERM	ECTODERM
Epithelial lining of gastrointestinal tract (except oral cavity and anal canal) and epithelium of its glands. Epithelial lining of urinary bladder, gallbladder, and liver. Epithelial lining of pharynx, auditory (eustachian) tubes, tonsils, tympanic (middle ear) cavity, larynx, trachea, bronchi, and lungs. Epithelium of thyroid gland, parathyroid glands, pancreas, and thymus. Epithelial lining of prostate and bulbourethral (Cowper's) glands, vagina, vestibule, urethra, and associated glands such as greater (Bartholin's) vestibular glands and lesser vestibular glands. Gametes (sperm and oocytes).	All skeletal and cardiac muscle tissue and most smooth muscle tissue. Cartilage, bone, and other connective tissues. Blood, red bone marrow, and lymphatic tissue. Blood vessels and lymphatic vessels. Dermis of skin. Fibrous tunic and vascular tunic of eye. Mesothelium of thoracic, abdominal, and pelvic cavities. Kidneys and ureters. Adrenal cortex. Gonads and genital ducts (except germ cells). Dura mater.	All nervous tissue. Epidermis of skin. Hair follicles, arrector pili muscles, nails, epithelium of skin glands (sebaceous and sudoriferous), and mammary glands. Lens, cornea, and internal eye muscles. Internal and external ear. Neuroepithelium of sense organs. Epithelium of oral cavity, nasal cavity, paranasal sinuses, salivary glands, and anal canal. Epithelium of pineal gland, pituitary gland, and adrenal medulla. Melanocytes (pigment cells). Almost all skeletal and connective tissue components of head. Arachnoid mater and pia mater.

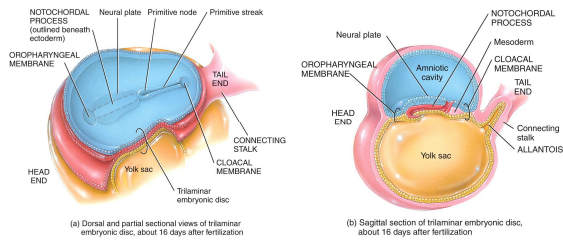
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Embryonic Period

- About 16 days after fertilization, the **notochordal process** forms.
- By days 22–24, the process becomes the solid cylinder called the **notochord**.
- The notochord is important for **induction**, the process whereby the **inducing tissue** stimulates development of a **responding tissue** to develop into a specific structure.
- The notochord induces the development of **vertebral bodies** and the **nucleus pulposus of vertebral discs**.

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Embryonic Period



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Embryonic Period

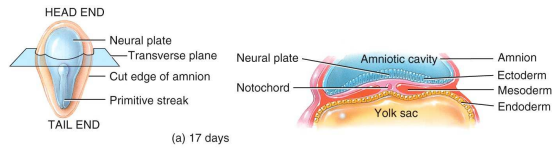
Also during the 3rd week of development, the following structures form:

- Oropharyngeal membrane
- Cloacal membrane
- Allantois

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Embryonic Period

The notochord also induces development of the **neural plate**.

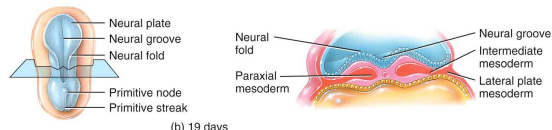


(a) 17 days

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Embryonic Period

The plate develops the **neural fold** as the lateral edges become more elevated.

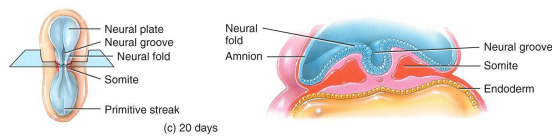


(b) 19 days

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Embryonic Period

The depressed midregion of the fold is the **neural groove**

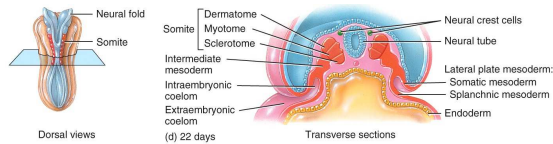


(c) 20 days

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Embryonic Period

As the neural folds approach each other and fuse, the **neural tube** is formed. The process for the formation of all of these structures is **neurulation**.



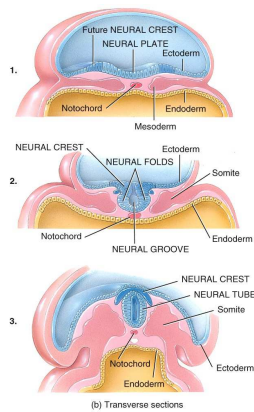
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Embryonic Period

As the neural tube forms, some of the ectodermal cells from the tube migrate to form several layers of cells called the **neural crest**.

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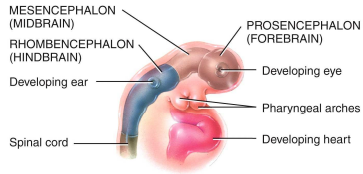
Embryonic Period



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Embryonic Period

At about 4 weeks after fertilization, the head end of the neural tube develops into three enlarged areas called **primary brain vesicles**.

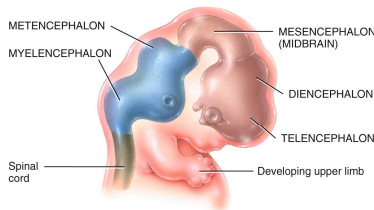


Lateral view of right side
(a) Three- to four-week embryo showing primary brain vesicles

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Embryonic Period

The vesicles are called the: **prosencephalon** (forebrain), **mesencephalon** (midbrain) and **rhombencephalon** (hindbrain).

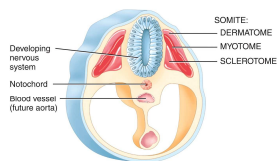


(b) Seven-week embryo showing secondary brain vesicles

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Embryonic Period

- By about the 17th day after fertilization, paired, cube-shaped structures called **somites** form. By the end of the 5th week, 42–44 pairs are present.
- Each somite differentiates into a **myotome**, a **dermatome** and a **sclerotome**.



(b) Transverse section through a somite showing its subdivisions

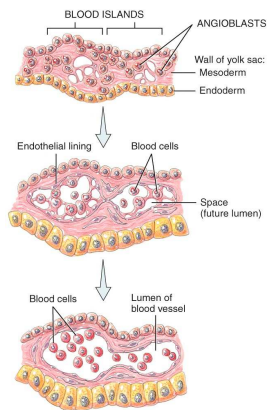
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Embryonic Period

At the beginning of the 3rd week, the **formation of blood vessels (angiogenesis)** begins with the development of **blood islands**.

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Embryonic Period



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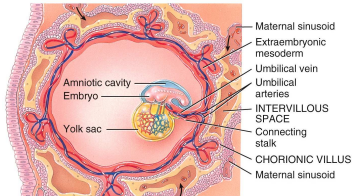
Embryonic Period

- On days 18 and 19, the heart begins to develop in the head end of the embryo. It begins in a region of **mesodermal cells** called the **cardiogenic area**.
- A pair of **endocardial tubes** forms.
- The tubes fuse to form a **primitive heart tube**.

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Embryonic Period

The vessels connecting to the heart do so by way of the **umbilical arteries** and **umbilical vein** through the body stalk which eventually becomes the **umbilical cord**.



Embryonic Period

- **Placentation** is the process of forming the **placenta**. This structure is the site of **exchange of nutrients and wastes between the mother and fetus**.
- The placenta produces hormones used to sustain the pregnancy.

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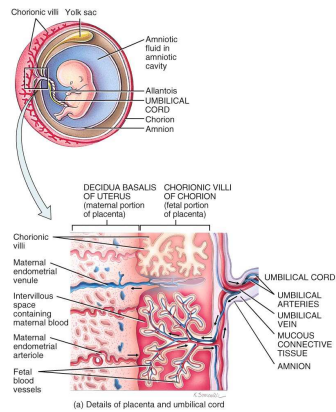
Embryonic Period

By the beginning of the 12th week, the placenta has two parts:

1. The **fetal portion** (chorionic villi)
2. The **maternal portion** (decidua basalis of the endometrium)

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Embryonic Period

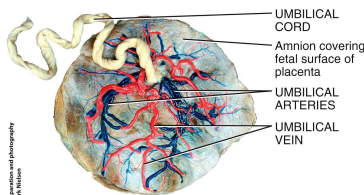


(a) Details of placenta and umbilical cord

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Embryonic Period

When fully developed, the placenta is shaped like a pancake. It is able to **protect the fetus from microorganisms** as well as its other functions.



(b) Fetal surface of placenta

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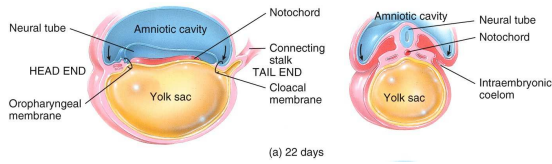
Embryonic Period

- All **major organs** develop between the **4th through 8th weeks (organogenesis)**.
- **Embryonic folding** occurs during the 4th week. This involves the flat embryo folding into a **three-dimensional cylinder**.

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Embryonic Period

A head fold and a tail fold develop.

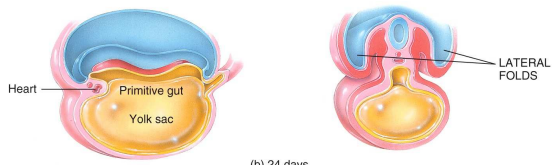


(a) 22 days

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Embryonic Period

Lateral folds form and as they move toward the midline they incorporate the **yolk sac** into the embryo as the **primitive gut**.

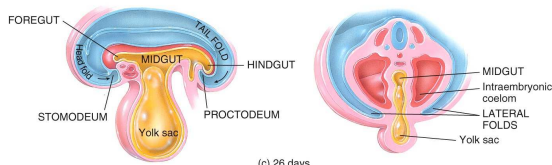


(b) 24 days

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Embryonic Period

On the outside of the embryo is a cavity in the tail region called the **proctodeum**.

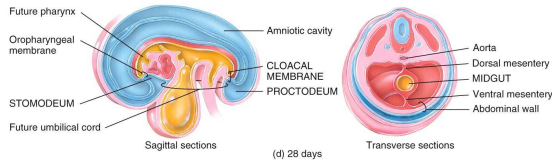


(c) 26 days

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Embryonic Period

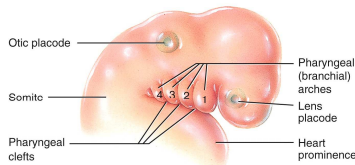
Separating the cloaca from the proctodeum is the **cloacal membrane**.



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Embryonic Period

Five pairs of **pharyngeal arches (branchial arches)** also develop on each side of the **future head and neck regions** during the 4th week. Each arch is separated by a **pharyngeal cleft**.

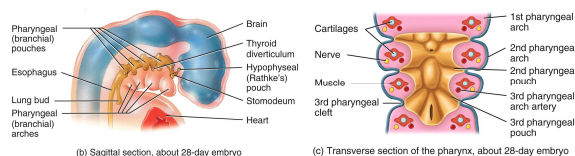


(a) External view, about 28-day embryo

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Embryonic Period

Pharyngeal pouches meet the pharyngeal clefts.



(b) Sagittal section, about 28-day embryo

(c) Transverse section of the pharynx, about 28-day embryo

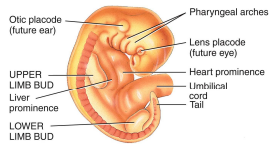
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Embryonic Period

By the middle of the 4th week, **upper limb buds** begin to develop.

By the end of the 4th week, **lower limb buds** and the **heart prominence** form.

At the end of the 4th week, the embryo has a **tail**.

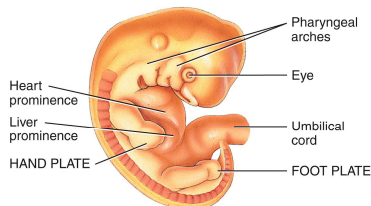


(b) Four-week embryo showing development of free limb buds

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Embryonic Period

During the 5th week, the **brain and head** develop rapidly and the **limbs develop further**.

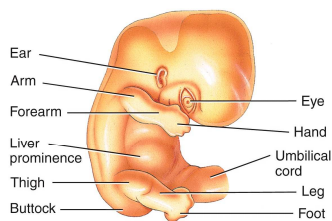


(c) Six-week embryo showing development of hand and foot plates

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Embryonic Period

By the 7th week, the regions of the limbs become distinct and **digits appear**.



(d) Seven-week embryo showing development of arm, forearm, and hand in free upper limb bud and thigh, leg, and foot in free lower limb bud

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Fertilization and Development

Interactions Animation:

- [Fertilization and Development](#)

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Teratogens

- Any agent or influence that is able to cause developmental defects in an embryo or fetus is a **teratogen**.
- Any number of **chemicals and drugs** may be considered teratogens. **Alcohol** is the most common (**fetal alcohol syndrome**).
- Others include **viruses, industrial chemicals, some hormones, antibiotics, cocaine** and **many others**.

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Teratogens

- **Cigarette smoking during pregnancy** has also been implicated as a cause of **low infant birth weight, cardiac abnormalities, anencephaly and higher infant and fetal mortality rates**.
- **Ionizing radiation** in many forms is also teratogenic. Exposure of the mother to **x-rays or radioactive isotopes** during pregnancy may cause **microcephaly** (small head), **mental retardation** and **skeletal deformities**.

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Prenatal Diagnostic Tests

- During pregnancy, several medical tests are used to detect **fetal abnormalities, genetic disorders** and **well-being**.
- **Fetal ultrasonography** is used to **determine a more accurate fetal age** when the date of conception is in doubt.
- It is also used to **confirm pregnancy, determine fetal position, identify multiple pregnancies** and other uses.

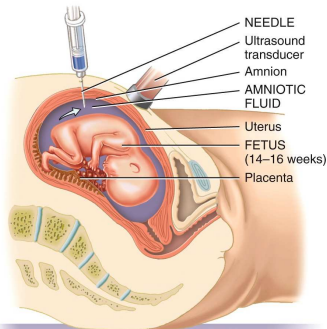
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Prenatal Diagnostic Tests

- **Amniocentesis** involves removing some **amniotic fluid** surrounding the developing fetus and analyzing it and fetal cells for **genetic abnormalities**. It is usually performed between **14–18 weeks**.
- The needle used to collect the fluid is guided by **ultrasound** to avoid damage to the fetus or umbilical cord.

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Prenatal Diagnostic Tests



(a) Amniocentesis

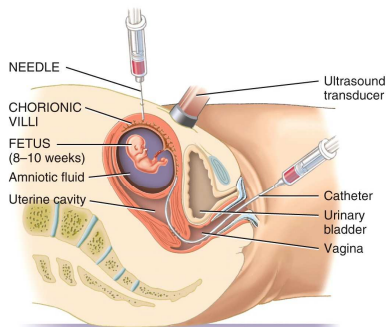
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Prenatal Diagnostic Tests

- **Chorionic villus sampling** may be performed as early as **8 weeks of gestation**.
- It is also done under ultrasound guidance, but the usual procedure is to insert a **catheter through the vagina and cervix** to collect a tissue sample from the chorionic villi.
- The goal is to identify the same genetic defects as seen with amniocentesis.
- The procedure may be done through the abdominal wall as with amniocentesis.

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Prenatal Diagnostic Tests



(b) Chorionic villi sampling (CVS)

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Prenatal Diagnostic Tests

- **Noninvasive prenatal tests** may also be performed, but they are currently not as informative as amniocentesis and chorionic villus sampling.
- The **maternal alpha-fetoprotein (AFP) test** requires a blood sample from the mother. It is used to detect AFP (a protein produced by the fetus at its highest levels between weeks 12-15) after the **16th week of pregnancy** when levels go to zero. High levels at this point indicate a **neural tube defect**.

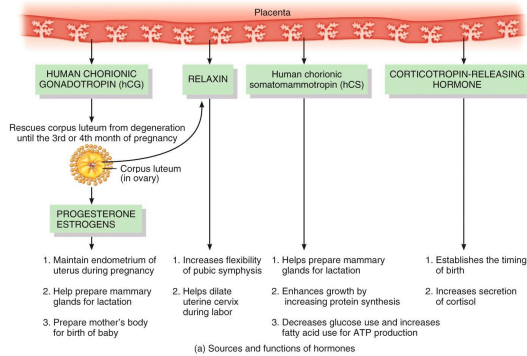
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Maternal Changes During Pregnancy

- During the first 3 to 4 months of pregnancy, the **corpus luteum** secretes **progesterone and estrogens in low levels**.
- From the 3rd month to the end of the pregnancy, the **placenta** produces high levels of these hormones.
- The **chorion** secretes **human chorionic gonadotropin (hCG)** to stimulate the corpus luteum to produce estrogens and progesterone to **inhibit menstruation** until the placenta takes over.

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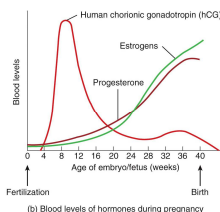
Maternal Changes During Pregnancy



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Maternal Changes During Pregnancy

- **hCG levels peak** at about the **9th week of pregnancy**.
- The **chorion** secretes **estrogens** after the first **3 or 4 weeks of pregnancy** and **progesterone** by the **6th week**



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Maternal Changes During Pregnancy

- **Relaxin** is secreted by the **corpus luteum** and later by the **placenta**. It **increases flexibility of the pubic symphysis and ligaments of the sacroiliac and sacrococcygeal joints** and also **helps dilate cervix during labor**.
- **Human chorionic somatomammotropin (hCS)**, also known as **human placental lactogen (hPL)**, probably helps prepare the mammary glands for lactation, helps maternal growth and regulates metabolism in mother and fetus.

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Maternal Changes During Pregnancy

- The hormone recently discovered to be secreted by the placenta is **corticotropin-releasing hormone (CRH)**. It is secreted in nonpregnant people by the hypothalamus. It is involved in the **timing of birth**.
- CRH is also needed to **increase secretion of cortisol** which is needed for **maturation of fetal lungs** and **production of surfactant**.

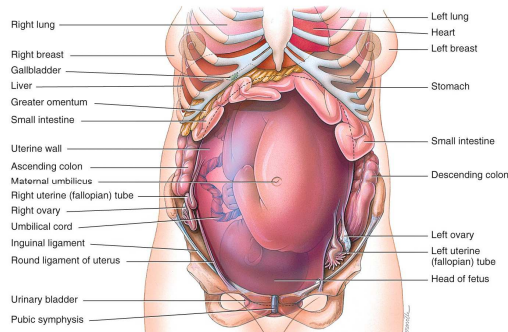
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Maternal Changes During Pregnancy

- The uterus continues to expand throughout the pregnancy moving upward into the abdominal cavity until it almost fills it.
- The organs are pushed out of the way and pressure on the stomach may cause food to be displaced causing heartburn.

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Maternal Changes During Pregnancy



Hormonal Regulation of Pregnancy and Childbirth

Interactions Animation:

- [Hormonal Regulation of Pregnancy and Childbirth](#)

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Exercise and Pregnancy

- Different factors during pregnancy may interfere with the ability to exercise.
- In early pregnancy, the mother **tires easily** and may suffer from **morning sickness**.
- **Weight increases and posture changes** as the pregnancy continues.
- Increased **relaxin** levels cause a change in gait.

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Labor

- **Labor** is the process that **expels the fetus from the uterus through the vagina.**
- Labor is initiated by the interaction of several hormones.
- Control of contractions occurs via a **positive feedback cycle.**

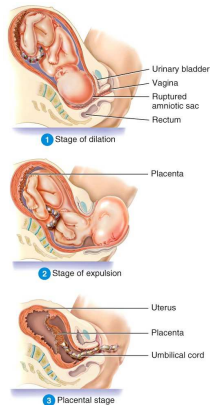
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Labor

- **True labor** begins when uterine contractions occur at regular intervals.
- **False labor** is associated with irregular contractions and no “show” (a discharge of blood with mucus).
- True labor is divided into three stages:
 1. Stage of dilation
 2. Stage of expulsion
 3. Placental stage

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Labor



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Labor

- Following delivery, it takes about 6 weeks for the maternal reproductive organs and physiology to return to the prepregnancy state. This period is the **puerperium**.
- The reduction in size of the uterus is **involution**.

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Adjustments of the Infant at Birth

- During development, the baby is totally dependent on the mother for survival.
- At birth, the fully developed newborn body begins to function independently.
- At birth, the lungs are able to exchange oxygen and carbon dioxide thanks to **surfactant** that began to develop by the end of the **6th month**.
- The **respiratory rate** at birth is **45 breaths per minute**, dropping to the normal **12 breaths per minute** within 2 weeks.

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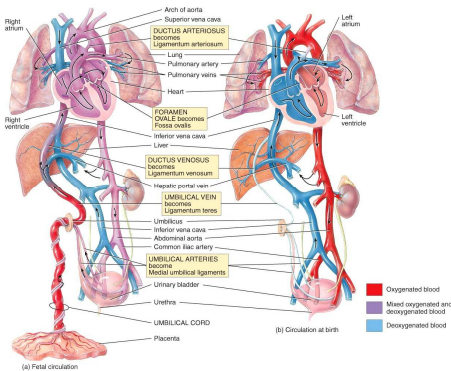
Adjustments of the Infant at Birth

After the baby's first breath, many changes must be made in the cardiovascular system over time.

- The **foramen ovale** closes to become the **fossa ovalis**.
- The **ductus arteriosus** closes to become the **ligamentum arteriosum**.
- The **umbilical arteries** fill with **connective tissue**.
- The **umbilical vein** becomes the **ligamentum teres of the liver**.

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Adjustments of the Infant at Birth



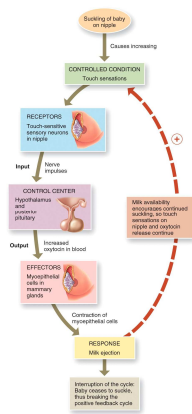
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The Physiology of Lactation

- **Lactation** is the production and ejection of milk from the **mammary glands**.
- **Prolactin (PRL)** (secreted by the anterior pituitary gland) is the main hormone in stimulating milk production.
- **Oxytocin** causes release of milk into the mammary ducts via the **milk ejection reflex**.

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The Physiology of Lactation



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The Physiology of Lactation

- There are benefits associated with breast feeding an infant:
 - The chemical composition of mother's milk is ideal for the baby's brain development, growth and digestion.
 - Several types of white blood cells (for immunity) are in the milk.
 - Antibodies are present.
 - Breast feeding supports optimal infant growth.
 - Breast feeding leads to a reduction in several diseases.

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Inheritance

- **Inheritance** is the passage of hereditary traits from one generation to the next. **Genetics** is the study of inheritance.
- Humans have **23 pairs of homologous chromosomes**; one in each pair from the father and one from the mother.
- Genes for the same trait that are in the same location on each homologue are **alleles**.
- A **mutation** is a permanent heritable change in an allele.

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Inheritance

- One genetic disorder caused by a mutation is **phenylketonuria (PKU)**.
- People with PKU cannot make the enzyme **phenylalanine hydroxylase** which is needed to break down phenylalanine.
- A **Punnett square** is used to show the possible genes inherited from two parents.

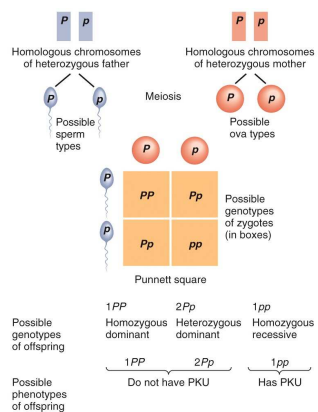
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Inheritance

- The **genotype** is the actual genetic makeup relating to a trait.
- An allele that dominates or masks the presence of another allele is a **dominant allele** (represented by an upper case letter)
- The allele whose presence is completely masked is the **recessive allele** (represented by a lower case letter).
- **Phenotype** is the physical expression of the genotype.

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Inheritance



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Inheritance

TABLE 29.3

Selected Hereditary Traits in Humans

DOMINANT	RECESSIVE
Normal skin pigmentation	Albinism
Near- or farsightedness	Normal vision
PTC taster*	PTC nontaster
Polydactyly (extra digits)	Normal digits
Brachydactyly (short digits)	Normal digits
Syndactylism (webbed digits)	Normal digits
Diabetes insipidus	Normal urine excretion
Huntington disease	Normal nervous system
Widow's peak	Straight hairline
Curved (hyperextended) thumb	Straight thumb
Normal Cl ⁻ transport	Cystic fibrosis
Hypercholesterolemia (familial)	Normal cholesterol level

*Ability to taste a chemical compound called phenylthiocarbamide (PTC).

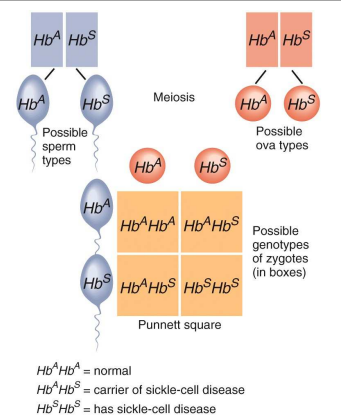
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Inheritance

- Most patterns of inheritance don't conform to the simple **dominant-recessive inheritance pattern**.
- **Incomplete dominance** is a situation where neither member of the pair of alleles is dominant over the other.
- An example of incomplete dominance is the inheritance of **sickle cell anemia**.

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Inheritance



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Inheritance

- **Multiple-allele inheritance** occurs when genes have more than two alternative forms.
- Inheritance of the **ABO blood group** is an example of this.
- Within this inheritance pattern there is also **codominance**. In this case, two genes (type A and type B blood) are expressed equally.

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Inheritance

Parents	A A	A B	A AB	A O	B B
Offspring	A, O	A, B, AB, O	A, B, AB	A, O	B, O

Parents	B AB	B O	AB AB	AB O	O O
Offspring	A, B, AB	B, O	A, B, AB	A, B	O

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Inheritance

- **Polygenic inheritance** is seen when a trait is controlled by the **combined effects of two or more genes**.
- **Complex inheritance** is seen when a trait occurs due to the **combined effects of many genes and environmental factors**.

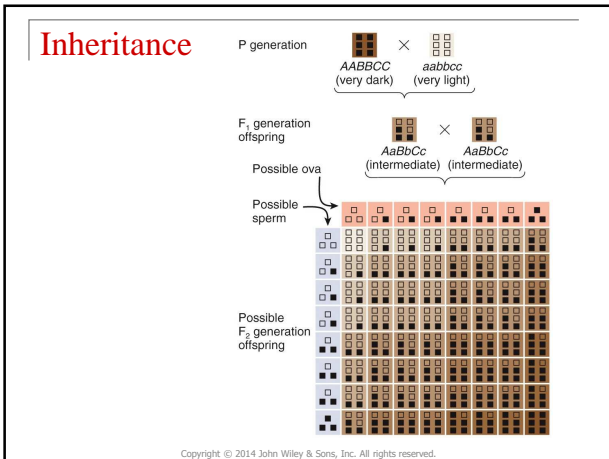
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Inheritance

Examples of complex traits include:

- Skin color
- Hair color
- Eye color
- Height
- Metabolic rate
- Body build

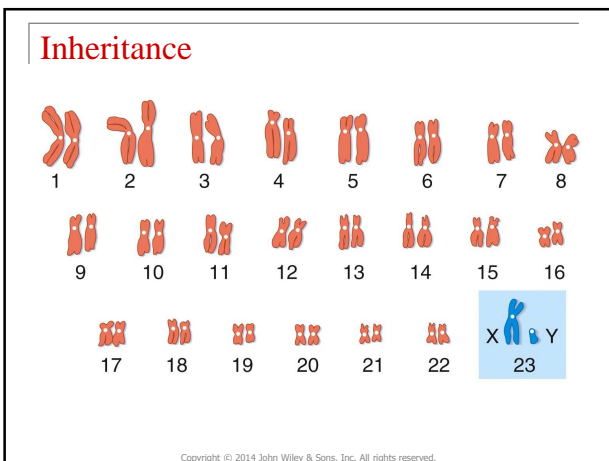
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Inheritance

- The 46 human chromosomes (23 pairs) are identified by their **size, shape and staining pattern**.
- An entire set of **chromosomes arranged in decreasing size order** and according to the position of the **centromere**, is called a **karyotype**.

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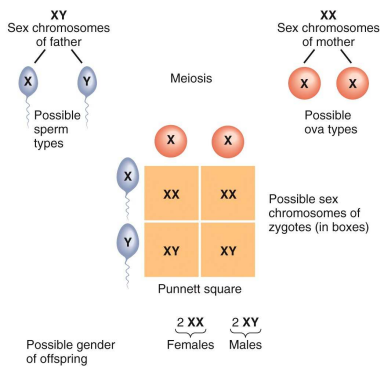


Inheritance

- The 23 pairs of human chromosomes include **22 pairs of autosomes** and **one pair of sex chromosomes (X and Y)**.
- Males have an **X and a Y chromosome**.
- Females have **two X chromosomes** (one is automatically inactivated—**X-chromosome inactivation**—and becomes a **Barr body**).
- Whether the sperm that will fertilize an egg is carrying an X or a Y chromosome will determine the gender of the zygote. An egg will only have one X chromosome under normal circumstances.

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Inheritance

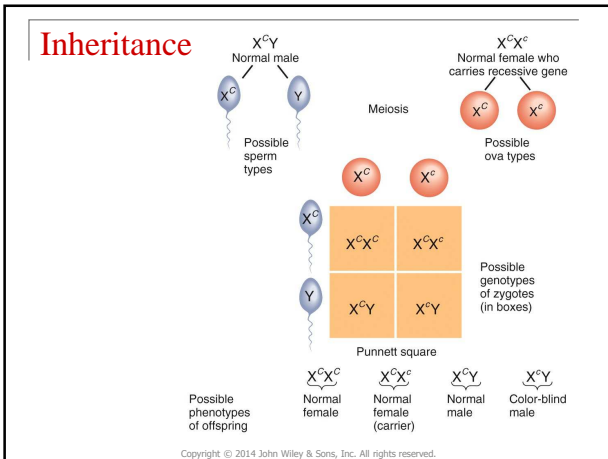


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Inheritance

- Some non-sexual traits are inherited on the X chromosome. These are called **sex-linked traits**.
- **Red-green color blindness** is an example of a sex-linked trait.

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End of Chapter 29

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