Chapter 29: Development and Inheritance
First week of development

Fertilization

- Genetic material from haploid sperm and haploid secondary oocyte merges into single diploid zygote
- Normally occurs in uterine (fallopian) tubes
- Sperm undergo capacitation – series of functional changes that prepare its plasma membrane to fuse with oocyte’s
- Sperm must penetrate coronoa radiata (granulosa cells) and zona pellucida (clear glycoprotein layer between corona radiate and oocyte plasma membrane)
- Acrosomal enzymes and strong movements help with penetration
Selected structures and events in fertilization

(a) Sperm cell penetrating a secondary oocyte

(b) Sperm cell in contact with a secondary oocyte

(c) Male and female pronuclei
Fusion of sperm cell and oocyte sets in motion events to block polyspermy – fertilization by more than one sperm

- Fast block to polyspermy – oocyte cell membrane depolarizes so another sperm cannot fuse
  - Also triggers exocytosis of secretory vesicles
- Slow block to polyspermy – molecules released in exocytosis harden entire zona pellucida

Oocyte must complete meiosis

- Divides into ovum and polar body (disintegrates)
First week of development (cont.)

- Male pronucleus and female pronucleus fuse to form single diploid (2n) zygote with 46 chromosomes
- Dizygotic (fraternal) twins are produced by the release of 2 secondary oocytes and fertilization by separate sperm
  - As genetically dissimilar as any other siblings
- Monozygotic (identical) twins develop form a single fertilized ovum – they have exactly the same DNA
  - Late separation results in conjoined twins
First week of development (cont.)

- **Cleavage of zygote**
  - Rapid mitotic cell divisions after zygote forms
  - First division begins 24 hours after fertilization and takes 6 hours
  - Each succeeding division takes less time
  - Blastomeres – progressively smaller cells produced by cleavage
  - Morula – solid sphere of cells
    - Still surrounded by zona pellucida
    - About same size as original zygote
First week of development (cont.)

- Blastocyst formation
  - Morula moves through uterine tubes toward uterus
  - Day 4 or 5 reaches uterus
  - Uterine milk – glycogen-rich secretions of endometrial glands nourishes morula
  - Blastocyst – at 32-cell stage, fluid collects and forms blastocyst cavity or blastocoel
  - 2 distinct cell populations
    - Embryoblast or inner cell mass – develops into embryo
    - Trophoblast – outer layer that forms wall and will ultimately develop into outer chorionic sac surrounding fetus and fetal portion of placenta
  - Day 5 “hatches” from zona pellucida
Cleavage and the formation of the morula and blastocyst

(a) Cleavage of zygote, two-cell stage (day 1)
(b) Cleavage, four-cell stage (day 2)
(c) Morula (day 4)
(d) Blastocyst, external view (day 5)
(e) Blastocyst, internal view (day 5)

Figure 29.02 Tortora - PAP 12/e
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Implantation

- About 6 days after fertilization attaches to the endometrium
- Orients inner cell mass toward endometrium
- 7 days after fertilization attaches more firmly and burrows in
  - Endometrium becomes more vascularized and glands enlarge
- Decidua – modified portion of endometrium after implantation
  - Regions named relative to site of implantation
Relationship of a blastocyst to the endometrium of the uterus at implantation

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

(b) Frontal section through endometrium of uterus and blastocyst, about 6 days after fertilization
Summary of events associated with the first week of development

1. Fertilization (occurs within uterine tube 12–24 hours after ovulation)
2. Cleavage (first cleavage completed about 30 hours after fertilization)
3. Morula (3–4 days after fertilization)
4. Blastocyst (4½–5 days after fertilization)
5. Implantation (occurs about 6 days after fertilization)

Frontal section through uterus, uterine tube, and ovary

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Second week of development

- Development of trophoblast
  - About 8 days after fertilization, trophoblast develops into 2 layers in region of contact between blastocyst and endometrium
    - Become part of chorion
  - Blastocyst becomes buried in endometrium and inner 1/3 of myometrium
  - Secretes human chorionic gonadotropin (hCG) that maintains corpus luteum so it continues to secrete estrogens and progesterone
    - Maintains uterine lining
Second week of development (cont.)

- Development of bilaminar embryonic disc
  - Cells of embryoblast also differentiate into 2 layers around 8 days after fertilization
    - Hypoblast (primitive endoderm)
    - Epiblast (primitive ectoderm)
      - Small cavity enlarges to form amniotic cavity
  - Development of amnion
    - Amnion forms roof of amniotic cavity and epiblast forms floor
    - Amnion eventually surrounds entire embryo
      - Amniotic cavity filled with amniotic fluid
      - Fluid derived from maternal blood and later fetal urine
Principal events in the second week of development
Development of yolk sac
- Also on 8th day after fertilization, cells at edge of hypoblast migrate to cover inner surface of blastocyst wall
- Form exocoelomic membrane
- Yolk sac – hypoblast and exocoelomic membrane
  - Relatively small and empty since nutrition derived from endometrium
  - Several important functions – supplies early nutrients, source of blood cells, contains primordial germ cells that migrate to gonads to form gametes, forms part of gut, functions as shock absorber, prevents desiccation
Second week of development (cont.)

- Development of sinusoids
  - 9th day after fertilization, blastocyst completely embedded in endometrium
  - Syncytiotrophoblast expands and spaces (lacunae) develop
  - 12th day – lacunae fuse to form lacunar networks
  - Endometrial capillaries dilate to form maternal sinusoids

- Development of extraembryonic coelom - about 12th day after fertilization
  - Fuse to form single large cavity

- Development of chorion
  - Formed by extraembryonic mesoderm and 2 layers or trophoblast
  - Becomes principal embryonic part of placenta
  - Protect embryo from immune responses of mother
  - Produces hCG
  - Connecting (body) stalk connects bilaminar embryonic disc to trophoblast – will become umbilical cord
Third week of development

- Begins 6 week period of rapid development and differentiation

- Gastrulation
  - 1st major event of 3rd week – about 15 days
  - Bilaminar embryonic disc transforms into trilaminar embryonic disc
    - Ectoderm (skin and nervous system), mesoderm (muscle, bones, connective tissues, peritoneum), and endoderm (epithelial lining of GI tract, respiratory tract, and several other organs)
    - Involves rearrangement and migration of epiblast cells
    - Primitive streak establishes head (primitive node) and tail ends
Gastrulation

(a) Dorsal and partial sectional views of embryonic disc, about 15 days after fertilization

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

- 16 days after fertilization notochord forms – induces tissue to become vertebral bodies
- 2 depressions form
  - Oropharyngeal membrane will later break down to connect mouth to pharynx and GI tract
  - Cloacal membrane will later degenerate to form openings of anus, urinary and reproductive tracts
- When cloacal membrane appears, wall of yolk sac forms allantois
  - Extends into connecting stalk
  - In most other mammals used for gas exchange and waste removal – human placenta does this instead
  - Does function in early formation of blood and blood vessels and urinary bladder
Development of the notochordal process

(a) Dorsal and partial sectional views of trilaminar embryonic disc, about 16 days after fertilization

(b) Sagittal section of trilaminar embryonic disc, about 16 days after fertilization
Third week of development (cont.)

- **Neurulation**
  - Notochord also induces formation of neural plate
  - Edges of plate elevate to form neural fold
  - Neural folds fuse to form neural tube
  - Develop into brain and spinal cord
  - Neural crest cells give rise to spinal and cranial nerves and ganglia, autonomic nervous system ganglia, CNS meninges, adrenal medullae and several skeletal and muscular components of head

- **Head end of neural tube develops into 3 primary brain vesicles**
  - Prosencephalon (forebrain), mesencephalon (midbrain), and rhombencephalon (hindbrain)
Third week of development (cont.)

- Development of somites
  - Mesoderm adjacent to notochord and neural tube forms paired longitudinal columns of paraxial mesoderm
  - Segment into paired, cube-shaped somites
  - Number of somites can be correlated to age of embryo
  - Each somite has 3 regions
    - Myotome – develops into skeletal muscles of neck, trunk and limbs
    - Dermatome – develops into connective tissue
    - Sclerotome - develops into vertebrae and ribs

- Development of intraembryonic coelom
  - Splits lateral plate mesoderm into
    - Splanchnic mesoderm – forms heart, blood vessels, smooth muscle and connective tissues of respiratory and digestive systems
    - Somatic mesoderm – gives rise to bones, ligaments, dermis of skin
Neurulation and the development of somites

Figure 20.29 Tonton - PAP 12.6
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Third week of development (cont.)

- Development of cardiovascular system
  - Angiogenesis – formation of blood vessels
    - Spaces develop in blood islands to form lumens of blood vessels
  - Pluripotent stem cells form blood cells
  - By end of 3rd week, heart forms and begins to beat

- Development of chorionic villi and placenta
  - Chorionic villi – fingerlike projections of chorion projecting into endometrium
  - Blood vessels in chorionic villi connect to embryonic heart through body stalk (becomes umbilical cord)
  - Maternal and fetal blood do not mix – diffusion only
Development of chorionic villi
Placentation

- Process of forming placenta
  - By beginning of 12th week has 2 parts
    - Fetal portion formed by chorionic villi of chorion
    - Maternal portion formed by decidua basalis of endometrium
  - Functionally allows oxygen and nutrients to diffuse from maternal to fetal blood while carbon dioxide and wastes diffuse from fetal to maternal blood
  - Not a protective barrier – allows microorganisms, drugs, alcohol to pass
  - Connection between embryo and placenta through umbilical cord
    - 2 umbilical arteries carry deoxygenated fetal blood to placenta
    - 1 umbilical vein carries oxygenated blood away from placenta
  - Afterbirth – placenta detaches from uterus
Placenta and umbilical cord

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Figure 29.11b Tortora - PAP 12/e
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Fourth week of development

- 4th-8th week - all major organs develop
- Organogenesis – formation of body organs and systems
- Embryo triples in size this week
- Converted from flat disc to 3D cylinder through embryonic folding
  - Main force is different rates of growth for different parts
- Head fold brings heart and mouth into eventual adult position
- Tail fold brings anus into eventual adult position
- Lateral folds for primitive gut – forerunner of GI tract
Embryonic folding

Figure 29.12 Tortora - PAP 12/e
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Somite and neural tube development

Pharyngeal (branchial) arches, clefts and pouches give rise to specific structures in head and neck

- 1st pharyngeal arch forms jaw

Otic placode – future internal ear

Upper and lower limb buds appear – distinct tail
5th – 8th weeks of development

- During 5th week brain develops rapidly so head growth considerable
- Limbs show substantial development by end of 6th week
  - Heart now 4-chambered
- 8th week
  - Digits of hands are short and webbed – by the end of the week the webbing dies (apoptosis)
  - Tail shorter and disappears by end of week
  - Eyes open – eyelids come together and may fuse
  - Auricles of ear visible
  - External genitals begin to differentiate
Fetal period

- During this period, tissues and organs developed during embryonic period grow and differentiate
- Very few new structures appear
- Rate of body growth remarkable
- Fetus less vulnerable to damaging effect of drugs, radiation, and microbes
## Summary of changes during embryonic and fetal development

### TABLE 29.2

<table>
<thead>
<tr>
<th>TIME</th>
<th>APPROXIMATE SIZE AND WEIGHT</th>
<th>REPRESENTATIVE CHANGES</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>EMBRYONIC PERIOD</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1–4 weeks</td>
<td>0.6 cm (3/16 in.) 0 g (0 oz)</td>
<td>Primary germ layers and notochord develop. Neurulation occurs. Primary brain vesicles, somites, and intraembryonic coeloms develop. Blood vessel formation begins and blood forms in yolk sac, allantois, and chorion. Heart forms and begins to beat. Chorionic villi develop and placental formation begins. The embryo folds. The primitive gut, pharyngeal arches, and limb buds develop. Eyes and ears begin to develop, tail forms, and body systems begin to form.</td>
</tr>
<tr>
<td>5–8 weeks</td>
<td>3 cm (1.25 in.) 1 g (1/30 oz)</td>
<td>Limbs become distinct and digits appear. Heart becomes four-chambered. Ears are far apart and eyelids are fused. Nose develops and is flat. Face is more humanlike. Bone formation begins. Blood cells start to form in liver. External genitalia begin to differentiate. Tail disappears. Major blood vessels form. Many internal organs continue to develop.</td>
</tr>
<tr>
<td><strong>FETAL PERIOD</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>9–12 weeks</td>
<td>7.5 cm (3 in.) 30 g (1 oz)</td>
<td>Head constitutes about half the length of the fetal body, and fetal length nearly doubles. Brain continues to enlarge. Face is broad, with eyes fully developed, closed, and widely separated. Nose develops a bridge. External ears develop and are low set. Bone formation continues. Upper limbs almost reach final relative length but lower limbs are not quite as well developed. Heartbeat can be detected. Gender is distinguishable from external genitalia. Urine secreted by fetus is added to amniotic fluid. Red bone marrow, thymus, and spleen participate in blood cell formation. Fetuses begin to move, but its movements cannot be felt yet by the mother. Body systems continue to develop.</td>
</tr>
<tr>
<td>13–16 weeks</td>
<td>18 cm (6.5–7 in.) 100 g (4 oz)</td>
<td>Head is relatively smaller than rest of body. Eyes move medially to their final positions, and ears move to their final positions on the sides of the head. Lower limbs lengthen. Fetuses appear more even humanlike. Rapid development of body systems occurs.</td>
</tr>
<tr>
<td>17–20 weeks</td>
<td>25–30 cm (10–12 in.) 200–450 g (0.5–1 lb)</td>
<td>Head is more proportionate to rest of body. Eyebrows and head hair are visible. Growth slows but lower limbs continue to lengthen. Vernix caseosa (fatty secretions of oil glands and dead epithelial cells) and lanugo (delicate fetal hair) cover fetus. Brown fat forms and is the site of heat production. Fetal movements are commonly felt by mother (quickening).</td>
</tr>
<tr>
<td>21–25 weeks</td>
<td>27–35 cm (11–14 in.) 550–800 g (1.25–1.5 lb)</td>
<td>Head becomes even more proportionate to rest of body. Weight gain is substantial, and skin is pink and wrinkled.</td>
</tr>
<tr>
<td>26–29 weeks</td>
<td>32–42 cm (13–17 in.) 110–1350 g (2.5–3 lb)</td>
<td>Head and body are more proportionate and eyes are open. Tonsils are visible. Body fat is 3.5% of total body mass and additional subcutaneous fat smooths out some wrinkles. Testes begin to descend toward scrotum at 28 to 32 weeks. Red bone marrow is major site of blood cell production. Many females born prematurely during this period survive if given intensive care because lungs can provide adequate ventilation and central nervous system is developed enough to control breathing and body temperature.</td>
</tr>
<tr>
<td>30–34 weeks</td>
<td>41–45 cm (16.5–18 in.) 2000–2300 g (4.5–5 lb)</td>
<td>Skin is pink and smooth. Fetus assumes upside-down position. Body fat is 8% of total body mass. Fetuses 33 weeks and older usually survive if born prematurely.</td>
</tr>
<tr>
<td>35–38 weeks</td>
<td>50 cm (20 in.) 3200–3400 g (7–7.5 lb)</td>
<td>By 38 weeks circumference of fetal abdomen is greater than that of head. Skin is usually bluish-pink, and growth slows as birth approaches. Body fat is 16% of total body mass. Testes are usually in scrotum in full-term male infants. Even after birth, an infant is not completely developed; an additional year is required, especially for complete development of the nervous system.</td>
</tr>
</tbody>
</table>

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*Table 29.2 from Tortora - PMP 12th Edition. All rights reserved.*
Summary of changes during embryonic and fetal development

Table 29.2 continued

| 4 | 8 | 12 | 16 | 20 | 24 | 28 | 32 | 36 (weeks) |

Table 29.03b: Torts + PAP 13e
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Summary of events of the embryonic and fetal periods

Figure 29.14  Tortora - PAP 12/e
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Maternal changes during pregnancy

- **Hormones of pregnancy**
  - 1\textsuperscript{st} 3-4 months of pregnancy, corpus luteum continues to secrete estrogens and progesterone
    - Maintains lining of uterus and prepares mammary glands to secrete milk
  - 3\textsuperscript{rd} month on, placenta produces high levels of estrogens and progesterone
  - Chorion secretes human chorionic gonadotropin (hCG)
    - Maintains corpus luteum
  - Relaxin – produced by corpus luteum and placenta
    - Increases flexibility of pubic symphysis
    - Helps dilate cervix during labor
Hormones during pregnancy

- Human chorionic somatomammotropin (hCS) or human placental lactogen (hPL) produced by chorion
  - Helps prepare mammary glands for lactation
  - Regulates certain aspects of fetal and maternal metabolism

- Corticotropin-releasing hormone (CRH) produced by placenta
  - In nonpregnant people secreted only by hypothalamus
  - Though to be part of “clock” establishing timing of birth
  - Increases secretion of cortisol needed for maturation of fetal lungs and production of surfactant
Hormones during pregnancy

(a) Sources and functions of hormones

1. Maintain endometrium of uterus during pregnancy
2. Help prepare mammary glands for lactation
3. Prepare mother's body for birth of baby

1. Increases flexibility of pubic symphysis
2. Helps dilate uterine cervix during labor

1. Helps prepare mammary glands for lactation
2. Enhances growth by increasing protein synthesis
3. Decreases glucose use and increases fatty acid use for ATP production

1. Establishes the timing of birth
2. Increases secretion of cortisol

(b) Blood levels of hormones during pregnancy

- Human chorionic gonadotropin (hCG)
- Estrogens
- Progesterone

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Changes during pregnancy

- By the end of a full-term pregnancy, uterus fills nearly the entire abdominal cavity
- Physiological changes
  - Weight gain due to fetus, amniotic fluid
  - Increased storage of proteins, triglycerides and minerals
  - Marked breast enlargement
  - Lower back pain – lordosis
- Changes in cardiovascular system due to increased maternal blood flow to placenta and increased metabolism
- Respiratory functions change to meet added oxygen demands of fetus
- Digestive system – increased appetite to meet energy demands of fetus
- Urinary system – pressure on bladder can cause incontinence
  - Increased renal filtering to eliminate wastes from fetus
Normal fetal location and position at the end of a full-term pregnancy
Labor or parturition

- Process by which fetus expelled from uterus through vagina
- Onset determined by interactions between several placental and fetal hormones
  - Levels of estrogen must rise to overcome inhibiting effect of progesterone on uterine contractions
  - High levels of estrogens increase number of receptors for oxytocin on uterine muscle fibers
  - Oxytocin stimulates contractions
  - Relaxin increases flexibility of pubic symphysis and dilates cervix
- Control of labor through positive feedback cycle
  - Contraction force fetal head into cervix which stretches
  - Stimulated stretch receptors cause release of more oxytocin
  - More oxytocin, more stretching
  - Cycle broken when stretching stops as baby exits
Stages of true labor

- True labor begins when uterine contractions occur at regular intervals
  - As interval shortens, contractions intensify
  - 3 stages
Adjustments of infant after birth

- **Respiratory adjustments**
  - Fetal lungs collapsed or partially filled with amniotic fluid
  - Respiratory system fairly well developed at least 2 months before birth
  - Rising CO$_2$ level in blood after delivery stimulates respiratory center in medulla oblongata causes respiratory muscle to contract
  - First inspiration is unusually deep with vigorous exhalation and crying

- **Cardiovascular adjustments**
  - Closure of foramen ovale between atria of fetal heart occurs at moment of birth
    - Diverts blood to lungs for the first time
    - Remnant called fossa ovalis
  - Ductus arteriosus constricts and becomes ligamentum arteriosum
    - Generally does not close completely for 3 months
  - Umbilical arteries become medial umbilical ligaments
  - Umbilical vein becomes round ligament of the liver
Physiology of lactation

- Secretion and ejection of milk from mammary glands
- Prolactin – principal hormone promoting milk synthesis and secretion
  - Secreted by anterior pituitary
  - Prolactin levels rise during pregnancy but progesterone inhibits effects of prolactin
  - After delivery, inhibition removed as estrogen and progesterone levels fall
  - Principal stimulus maintaining prolactin secretion is sucking action of infant
    - Impulses from stretch receptors decrease release of prolactin-inhibiting hormone (PIH) and increases release of prolactin-releasing hormone (PRH) from hypothalamus
The milk ejection reflex

- Oxytocin causes milk ejection reflex
  - Suckling, hearing baby cry, touching mother’s genitals can initiate
- Colostrum – before appearance of true milk on 4th day
  - Contain important antibodies
- Lactation often blocks ovarian cycles for few months after delivery
- Primary benefit of breast-feeding is nutritional
  - Other benefits also
Inheritance

- Passage of hereditary traits from one generation to the next

Genotype and phenotype

- Nuclei of all human cells except gametes contain 23 pairs of chromosomes – diploid or 2n
- One chromosome from each pair came from father, other member from mother
- Each chromosome contains homologous genes for same traits
- Allele – alternative forms of a gene that code for the same trait
- Mutation – permanent heritable change in allele that produces a different variant
Phenylketonuria or PKU example

- Unable to manufacture enzyme phenylalanine hydroxylase
- Allele for function enzyme = P
- Allele that fails to produce functional enzyme = p
- Punnet square show possible combinations of alleles between 2 parents
- Genotype – different combinations of genes
- Phenotype – expression of genetic makeup
  - PP – homozygous dominant – normal phenotype
  - Pp – heterozygous – normal phenotype
    - 1 dominant allele codes for enough enzyme
    - Can pass recessive allele on to offspring – carrier
  - pp - homozygous recessive – PKU
    - 2 recessive alleles make no functional enzyme
Inheritance

**TABLE 29.3**

Selected Hereditary Traits in Humans

<table>
<thead>
<tr>
<th>DOMINANT</th>
<th>RECESSIVE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal skin pigmentation</td>
<td>Albinism</td>
</tr>
<tr>
<td>Near- or farsightedness</td>
<td>Normal vision</td>
</tr>
<tr>
<td>PTC taster*</td>
<td>PTC nontaster</td>
</tr>
<tr>
<td>Polydactyly (extra digits)</td>
<td>Normal digits</td>
</tr>
<tr>
<td>Brachydyactyly (short digits)</td>
<td>Normal digits</td>
</tr>
<tr>
<td>Syndactylysm (webbed digits)</td>
<td>Normal digits</td>
</tr>
<tr>
<td>Diabetes insipidus</td>
<td>Normal urine excretion</td>
</tr>
<tr>
<td>Huntington disease</td>
<td>Normal nervous system</td>
</tr>
<tr>
<td>Widow’s peak</td>
<td>Straight hairline</td>
</tr>
<tr>
<td>Curved (hyperextended) thumb</td>
<td>Straight thumb</td>
</tr>
<tr>
<td>Normal Cl⁻ transport</td>
<td>Cystic fibrosis</td>
</tr>
<tr>
<td>Hypercholesterolemia (familial)</td>
<td>Normal cholesterol level</td>
</tr>
</tbody>
</table>

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

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Inheritance

- Alleles that code for normal traits are not always dominant
  - Huntington disease caused by dominant allele
    - Both homozygous dominant and heterozygous individuals get HD

- Nondisjunction
  - Error in cell division resulting in abnormal number of chromosomes
  - Aneuploid – chromosomes added or missing
    - Monosomic cell missing 1 chromosome (2n-1)
    - Trisomic cell has additional chromosome (2n +1)
      - Down Syndrome – trisomy 21 – 3 21st chromosomes
Variations of Dominant-recessive inheritance

- Simple dominance-recessive
  - Just described where dominant allele covers effect of recessive allele
- Incomplete dominance
  - Neither allele dominant over other
  - Heterozygote has intermediate phenotype
  - Sickle-cell disease
Sickle-cell disease

- **Sickle-cell disease**
  - $Hb^A Hb^A$ – normal hemoglobin
  - $Hb^S Hb^S$ – sickle-cell disease
  - $Hb^A Hb^S$ – ½ normal and ½ abnormal hemoglobin
  - Minor problems, are carriers for disease

![Diagram showing possible genotypes of zygotes](figure.png)

- $Hb^A Hb^A = normal$
- $Hb^A Hb^S = carrier of sickle-cell disease$
- $Hb^S Hb^S = has sickle-cell disease$

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Multiple-allele inheritance

- Some genes have more than 2 alleles
- ABO blood group
  - $I^A$ produces A antigen
  - $I^B$ produces B antigen
  - i produces neither
  - A and B are codominant
  - Both genes expressed equally in heterozygote

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype (blood type)</th>
</tr>
</thead>
<tbody>
<tr>
<td>$I^A I^A$ or $I^A i$</td>
<td>A</td>
</tr>
<tr>
<td>$I^B I^B$ or $I^B i$</td>
<td>B</td>
</tr>
<tr>
<td>$I^A I^B$</td>
<td>AB</td>
</tr>
<tr>
<td>i</td>
<td>O</td>
</tr>
</tbody>
</table>
Blood type inheritance

Parents

A  A  A  B  A  AB  A  O  B  B

A, O  A, B, AB, O  A, B, AB  A, O  B, O

Offspring

Parents

B  AB  B  O  AB  AB  AB  O  O  O

A, B, AB  B, O  A, B, AB  A, B  O

Offspring

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Complex inheritance

- Polygenic inheritance – most inherited traits not controlled by one gene
- Complex inheritance – combined effects of many genes and environmental factors
  - Skin color, hair color, height, metabolism rate, body build
  - Even if a person inherits several genes for tallness, full height can only be reached with adequate nutrition
  - Neural tube deficits are more common if the mother lacks adequate folic acid in the diet – environmental effect
Skin color is a complex trait

- Depends on environmental conditions like sun exposure and nutrition and several genes
- Additive effects of 3 genes plus environmental affect produces actual skin color
Autosomes, sex chromosomes and sex determination

- Karyotype shows 46 chromosomes arranged in pairs by size and centromere position
- 22 pairs are autosomes – same appearance in males and females
- 23rd pair are sex chromosomes
  - XX = female
  - XY = male
Sex determination

- Males produce sperm carrying an X or Y
  - Females only produce eggs carrying an X
  - Individual’s sex determined by father’s sperm carrying X or Y
- Male and female embryos develop identically until about 7 weeks
  - Y initiates male pattern of development
    - SRY on Y chromosome
  - Absence of Y determines female pattern of development

![Sex chromosomes of father and mother](image)

Possible sex chromosomes of zygotes (in boxes)

- Punnett square
  - 2 XX Females
  - 2 XY Males

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Sex-linked inheritance

- Genes for these traits on the X but not the Y
- Red-green colorblindness
  - Most common type of color blindness
  - Red and green are seen as same color
  - Males have only one X
    - They express whatever they inherit from their mother

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>$X^C_X^C$</td>
<td>Normal female</td>
</tr>
<tr>
<td>$X^C_X^c$</td>
<td>Normal female (carrier)</td>
</tr>
<tr>
<td>$X^c_X^c$</td>
<td>Color blind female</td>
</tr>
<tr>
<td>$X^C_Y$</td>
<td>Normal male</td>
</tr>
<tr>
<td>$X^c_Y$</td>
<td>Color blind male</td>
</tr>
</tbody>
</table>
Inheritance of red-green color blindness

Figure 29.26 Tortora - PAP 12/e
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