Chapter 29
Development and Inheritance

- From fertilization to birth
  - fertilization
  - implantation
  - placental development
  - fetal development
  - gestation
  - labor
  - parturition (birth)

Terminology of Development

- Gestation period
  - time span from fertilization to birth (38 weeks)
- Prenatal period (before birth)
  - embryological development
    - developing human for first 2 months after fertilization is known as an embryo
    - all principal adult organs are present
  - fetal development
    - from 9 weeks until birth is known as a fetus
    - by end of 3rd month, placenta is functioning
- Neonatal period is first 42 days after birth

From Fertilization to Implantation
Events Before Fertilization

- Haploid sperm nucleus & haploid secondary oocyte nucleus merge to form a single diploid nucleus
- Occurs in uterine tube within 24 hours after ovulation (oocyte dies in 24 hours)
- Events occurring before fertilization
  - peristalsis of uterine tube & movement of cilia transport the oocyte towards the uterus
    - oocyte releases chemical attractants
  - sperm swim towards oocyte by means of flagella
    - prostaglandins within the semen stimulate uterine contractions that help move sperm towards the oocyte
  - capacitation or final maturation of the sperm occurs within female
    - acrosomal membrane becomes fragile

Sperm Contact during Fertilization

- Sperm penetrates the granulosa cells around the oocyte (corona radiata)
- Sperm digests its way through the zona pellucida
  - when ZP3 glycoprotein binds to sperm head, it triggers the acrosomal reaction (enzyme release)
- First sperm to fuse with oocyte membrane triggers the slow & the fast block to polyspermy
  - 1-3 seconds after contact, oocyte membrane depolarizes & other cells can not fuse with it = fast block to polyspermy
  - depolarization triggers the intracellular release of Ca+2 causing the exocytosis of molecules hardening the entire zona pellucida = slow block to polyspermy

Events Within the Egg

- Sperm entry, triggers oocyte to complete meiosis II and dump second polar body
- Once inside the oocyte, the sperm loses its tail & becomes a male pronucleus
- Fusion of male & female haploid pronuclei is the true moment of fertilization
- Fertilized ovum (2n) is called a zygote
  - zona pellucida still surrounds it

Formation of the Morula

- Rapid mitotic cell division of embryo is called cleavage
- 1st cleavage in 30 hours produces 2 blastomeres
- 2nd cleavage on 2nd day
- By 3rd day has 16 cells
- By day 4 has formed a solid ball of cells called a morula

Development of the Blastocyst

- A blastocyst is a hollow ball of cells that enters the uterine cavity by day 5
  - outer covering of cells called the trophoblast
  - inner cell mass
  - fluid-filled cavity called the blastocele
- Trophoblast & part of inner cell mass will develop into fetal portion of placenta
- Most of inner cell mass will become embryo

Implantation

- Attachment of blastocyst to endometrium
  - occurs 6 days after fertilization
  - implants with inner cell mass in contact with the endometrium
- Trophoblast develops 2 distinct layers
  - syncytiotrophoblast secretes enzymes that digest the endometrial cells
  - cytotrophoblast is distinct layer of cells that defines the original shape of the embryo
- Trophoblast secretes human chorionic gonadotropin (hCG) that helps the corpus luteum maintain the uterine lining

Beginnings of Organ Systems(Gastrulation)

- Day 8
  - cytotrophoblast forms amnion & amnionic cavity
    - cells of inner cell mass on amnionic cavity form ectoderm
    - cells bordering on blastocele form endoderm
  - ectoderm & endoderm together form embryonic disk

- Day 12
  - endodermal cells divide to form a hollow sphere (yolk sac)
  - cytotrophoblast cells divide to fill the spaces surrounding the yolk sac with extraembryonic mesoderm
    - spaces develop in that layer to form future ventral body cavity

Primary Germ Layers

- Day 14 --cells of embryonic disc produce 3 distinct layers
  - endoderm forms epithelial lining of GI & respiratory
  - mesoderm forms muscle, bone & other connective tissues
  - ectoderm develops into epidermis of skin & nervous system

Formation of Embryonic Membranes

- Yolk sac
  - site of early blood formation
  - gives rise to gonadal stem cells (spermatogonia & oogonia)
- Amnion
  - surrounds embryo with fluid: shock absorber, regulates body temperature & prevents adhesions
  - fluid is filtrate of mother's blood + fetal urine
  - examine a sample of it for embryonic cells (amniocentesis)
- Chorion
  - becomes the embryonic contribution to the placenta
  - derived from trophoblast & mesoderm lining it

  - gives rise to human chorionic gonadotropin (hCG)
- Allantois
  - outpocketing off yolk sac that becomes umbilical cord

Placenta & Umbilical Cord

- Placenta forms during 3rd month
  - chorion of embryo & stratum functionalis layer of uterus
- Chorionic villi extend into maternal blood filled intervillous spaces --- maternal & fetal blood vessels do not join & blood does not mix
  - diffusion of O2, nutrients, wastes
  - stores nutrients & produces hormones
  - barrier to microorganisms, except some viruses
    - AIDS, measles, chickenpox, poliomyelitis, encephalitis
  - not a barrier to drugs such as alcohol
- Placenta detaches from the uterus (afterbirth)

Parts of Endometrial Lining

- Decidua = all of endometrium lost as placenta
  - equals all of the endometrium, except stratum basalis
- Decidua basalis---portion of endometrium deep to chorion
- Decidua capsularis---part of endometrial wall that covers implanted embryo
- Decidua parietalis---part of endometrial wall not modified by embryo until embryo bumps into it as it enlarges
- Decidua capsularis fuses with decidua parietalis

Umbilical Cord

- Contents
  - 2 arteries that carry blood to the placenta
  - 1 umbilical vein that carries oxygenated blood to the fetus
  - primitive connective tissue
- Stub drops off in 2 weeks leaving a scar called the umbilicus

Hormones of Pregnancy

- Chorion
  - from day 8 until 4 months secretes hCG which keeps corpus luteum active
  - corpus luteum produces progesterone & estrogen to maintain lining of uterus
- Placenta
  - by 4th month produces enough progesterone & estrogen that corpus luteum is no longer important
  - relaxin which relaxes CT of pelvis and cervix
  - human chorionic somatomammotropoin (hCS) or human placental lactogen (hPL)
    - maximum amount by 32 weeks
    - helps prepare mammary glands for lactation
  - corticotropin-releasing hormone (CRH) increases secretion of fetal cortisol (lung maturation) & acts to establish timing of birth

Hormone Blood Levels

- Human chorionic gonadotropin (hCG) produced by the chorion is less important after 4 months, because the placenta takes over the hormonal secretion of the corpus luteum.

Hormonal Secretion by the Placenta

Developmental Changes

- Read Table 29.2 to get a full description of the timing of fetal events during development

Maternal Changes During Pregnancy

- Uterus nearly fills the abdominal cavity
- GI tract compressed causing heartburn & constipation
- Pressure on bladder causing changes in frequency & urgency
- Compression of vena cava causing varicose veins & edema in the legs
- Compression of renal vessels causing renal hypertension
- Cardiovascular changes to meet needs of fetus
  - rise in cardiac output of 20-30% due to placenta
  - increase in heart rate 15% & increase in blood volume 30-50%
- Respiratory changes
  - increase in tidal volume 30%
  - decrease in expiratory reserve volume & airway resistance
  - minute respiratory volume increases as O2 needs increase
- Reproductive system changes
  - uterus increases in size from 80 g to 1200g
  - hyperplasia and hypertrophy
- Urinary system changes
  - increase in glomerular filtration rate of 40%

Labor and Parturition

- Parturition means giving birth; labor is the process of expelling the fetus
- Labor begins when progesterone's inhibition is overcome by an increase in the levels of estrogen
  - progesterone inhibits uterine contraction
  - placenta stimulates fetal anterior pituitary which causes fetal adrenal gland to secrete DHEA
  - placenta converts DHEA to estrogen
  - estrogen overcomes progesterone and labor begins

Positive Feedback during Labor

- Uterine contraction forces fetal head into cervix (stretch)
- Nerve impulses reach hypothalamus causing release of oxytocin
- Oxytocin causes more contractions producing more stretch of cervix & more nerve impulses

True Versus False Labor

- True labor begins when contractions occur at regular intervals
  - produces pain
    - back pain increases with walking
  - dilation of cervix with a discharge of blood-containing mucus in the cervical canal
- False labor produces pain at irregular intervals but there is no cervical dilation

Stages of Labor

- Dilation
  - 6 to 12 hours
  - rupture of amniotic sac & dilation of cervix
- Expulsion
  - 10 minutes to several hours
  - baby moves through birth canal
- Placental
  - 30 minutes
  - afterbirth is expelled by muscular contractions

Physiology of Lactation

- Lactation = production & release of milk
- Prolactin from anterior pituitary increases during pregnancy, but progesterone inhibits effects of prolactin until after delivery
- After delivery, progesterone levels drop & suckling increases the release of prolactin & oxytocin (milk ejection reflex)
- Colostrum = cloudy fluid released for few days
- True milk produced by 4th day

- If suckling stops, milk secretion stops

Milk Ejection Reflex

- Oxytocin cause release of milk into mammary ducts
- Stimulation of touching nipple causes hypothalamus to release oxytocin
- Oxytocin causes contraction of myoepithelial cells
- Milk moved from alveoli into mammary ducts
- Oxytocin release by other stimuli
  - hearing a baby's cry or touching the genitals

Inheritance

- Passing of hereditary traits from one generation to the next
- Genotype
  - all human cells contain 23 pairs of chromosomes
    - one chromosome in each pair came from the mother and the other came from the father
    - similar locations on each pair of chromosomes code for the same trait (alleles)
      - if one allele controls the express of a trait, it is the dominant allele
      - if the other allele is completely masked it is the recessive allele
    - a person with the same alleles on both chromosomes is said to be homozygous for the trait----heterozygous for the trait is having different alleles on homologous chromosomes
  - heterozygous individuals are carriers of a recessive gene

Genotype & Phenotype

- Genotype = your genetic makeup
- Phenotype = what you look like (outward expression of your genes)
- Punnett square
  - method of showing 4 possible genetic combinations in offspring of 2 individuals

Genetic Problems

- Error in meiosis called nondisjunction
  - chromosomes fail to separate properly
  - cell with one or more extra or missing chromosomes is called an aneuploid
    - (2n-1) is missing a chromosome
    - (2n+1) has an extra chromosome
- Error in meiosis called translocation
  - location of chromosome segment is moved
    - crossing-over between 2 nonhomologous chromosomes
    - Down syndrome results from a portion of chromosome 21 becoming part of another chromosome
      - individuals have 3 copies of that part of chromosome 21

Incomplete Dominance

- Neither member of an allelic pair is dominant over the other --- resulting phenotype is intermediate
- Sickle-cell trait individuals have both HbA & HbS
  - suffer from only minor problems with anemia since have both normal & sickle-cell hemoglobin
- Sickle-cell anemic individuals have 2HbS alleles
  - produce sickle-cell hemoglobin
  - suffer from severe anemia

Sickle-Cell Inheritance

- 1 normal
- 2 embryos will be sickle-cell trait
- 1 sickle-cell anemia

Multiple-Allele Inheritance

- Genes with more than two alternate forms
  - 3 different alleles of the I gene
  - I^A, I^B, or i
    - A and B alleles are codominant since both genes are expressed equally
    - 6 possible genotypes produce 4 blood types
  - 4 phenotypes of the ABO blood groups are (A, B, AB & O)

Polygenic Inheritance

- Traits controlled by many genes
  - continuous gradations of small differences
  - body build, height and skin, hair & eye color
- Skin color controlled by 3 genes (Aa, Bb, Cc)
  - person with genotype of AABBCC is dark
  - person aabbcc is light
- Parental generation & F1 and F2 generation

Autosomes & Sex Chromosomes

- Each of us has a pair of sex chromosomes
- Females XX
- Males have XY
  - Y is smaller
  - Y is needed to produce male development

Human Chromosomes

- 22 pairs of autosomes
- 1 pair of sex chromosomes

Sex-Linked Inheritance

- Genes found only on X chromosomes
- Red-Green color blindness is lack of either red or green cones, so seen as same color
  - X^CX^C is normal, X^CX^c is carrier
  - X^cX^c is color blind
  - X^CY is normal, X^cY is color blind
- Hemophilia is sex-linked trait where blood fails to clot
- Other sex-linked traits
  - absence of incisors, night blindness, juvenile glaucoma, and some types of deafness, diabetes, cataracts, and muscular dystrophy

X-Chromosome Inactivation

- Females have double dose of X chromosome in all cells
- One X chromosome is randomly & permanently inactivated early in development
- Visible as dark-staining Barr body easily seen in nucleus of neutrophils as “drumstick”
  - tightly coiled even in interphase cell

Environmental Influences

- Phenotype is result of environment effects on genetic makeup
  - more influential on polygenic traits such as height
- Teratogens = cause developmental defects
  - Chemicals & Drugs
    - fetal alcohol syndrome = slow growth, facial features, defective heart & CNS
    - cocaine = attention problems, hyperirritability, seizures
  - Cigarette Smoking
    - low birth weight, cleft lip & palate, SIDS
  - Irradiation or radioisotopes during first trimester
    - mental retardation, microcephaly