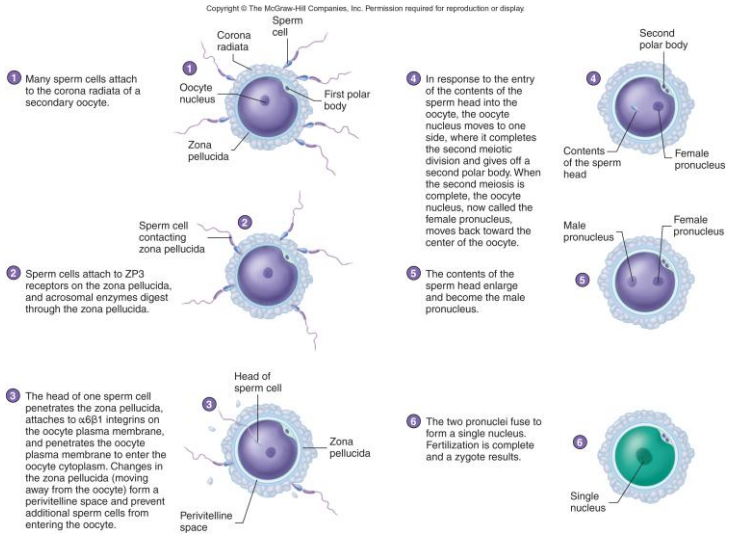
Unit 4 Lecture 13

PREGNANCY, GROWTH, & DEVELOPMENT

Development during Pregnancy

For fertilization to occur, a sperm must penetrate an ovum within 24 hours of ovulation. Therefore, fertilization has to happen in the distal end of the Fallopian tube. An unfertilized ovum dies by the time it reaches the uterus. Sperm can reach the distal end of the Fallopian tube within 5-10 minutes after ejaculation. However, they cannot fertilize the egg for another ten hours. The sperm need to undergo a process called capacitation; a process that makes it possible for the sperm to penetrate the egg.



The first sperm that reaches the egg is not the one to fertilize it. It may take a hundred sperm to invade the follicular cells, clear a path for the one sperm that binds to the *zona pellucida.* This one sperm releases enzymes digesting its way through the *zona* until it contacts the egg. The sperm head and midpiece enter the egg but the egg destroys paternal mitochondria and the tail of the sperm.

Fertilization of the ovum by a sperm cell results in the formation of a zygote. Once this happens additional sperm are prevented from penetrating the oocyte by fast and slow block mechanisms. Completion of female meiosis II occurs only after the sperm enters the egg. Even under optimum conditions, pregnancy only occurs about 25% of the time. Research indicates that about 50% of the fertilized zygotes are lost. Of the successful pregnancies, 20% will result in a miscarriage. Most of the miscarriages are due to chromosomal abnormalities in the zygote.

The period of cleavage is the first period in the prenatal period. Cleavage is defined as mitotic cellular divisions. The formation of the morula occurs about 3 days after fertilization, multiple cellular divisions and the arrival at the uterus. The zona pellucida disintegrates. Development of the blastocyst, a hollow ball of cells that implants in the endometrium, occurs about six days after fertilization begins. The blastocyst has an outer covering called a trophoblast that eventually develops into the chorion. The inner cell mass, called the embryoblast, develops into the embryo proper. It is during this time of early cellular division that the chance for multiple births occurs from the same egg. Whereas dizygotic (fraternal) twins are the result of independent release and fertilization of two oocytes, monozygotic (identical)

develop from a single fertilized ovum in which the embryoblast later divides into two.

Implantation occurs about six days after fertilization when the blastocyst buries itself in the endometrium. Trophoblastic cells of the blastocyst secrete enzymes that stimulate a thickening of the endometrium. A layer of cells from the trophoblast grows into the uterus while the uterus grows over the trophoblast, enclosing it. The trophoblast secretes the enzyme hCG.

Embryonic development occurs from the second week through the eighth week of development. The beginning of the organ systems occur during embryonic development. The primary germ layers are, initially the ectoderm and endoderm layers (form embryonic disc), and eventually the mesoderm which develops in between these two layers. The endoderm becomes endothelial lining of GI tract and other accessory organs. The mesoderm forms muscles, bone and other connective tissue and the peritoneum. The ectoderm develops into the skin and nervous tissue.

Embryonic membranes

The yolk sac is the primary source of nourishment to developing embryo and early source of blood development as a result of the embryo receiving nutrients from endometrium of uterus. The yolk sac contributes to the formation of the digestive tract and future eggs or sperm cells. The amnion becomes amniotic sac and is filled with amniotic fluid. This bag of waters helps protect the growing fetus from trauma and temperature fluctuations. The chorion becomes the fetal portion of the placenta. The allantois develops into to the blood vessels of the umbilical cord and part of the urinary bladder. The placenta and umbilical cord are expelled after birth. The **fetal stage** begins at the eighth week and terminates at birth.

During **gestation** the fetus grows and develops. There are some differences in systems prior to birth and after birth. The main ones concern the circulatory and the respiratory systems. The circulatory system in the fetus is slightly different than the one after birth. Prior to birth the maternal blood supply provides nutrients and oxygen and carries away wastes which diffuse thought the placenta. One half of the blood traveling into the fetus via the umbilical vein goes to the fetal liver while the other half goes via the *ductus venosus* to the inferior vena cava.

Since fetal lungs are nonfunctional, blood does not need to go to them. Instead, blood passes from the right atrium to the left atrium through the foramen ovale. From the heart most of the blood goes into the pulmonary trunk passes by way of the *ductus arteriosus* to the aorta to go to the rest of the body. The blood then returns via the iliac arteries to the placenta in umbilical arteries. The small amount of blood that does go to the lungs, returns to the heart and leaves the heart via the aorta.

At birth the foramen ovale closes by the closing of two flaps of tissue that fuse and becomes the fossa ovalis. The *ductus arteriosus* closes almost immediately and usually within three months after birth becoming the ligamentum arteriosum. The *ductus venosus* closes becoming the ligamentum venosum. The respiratory system in the fetus depends on mom for gas exchange, but at birth it immediately takes over by itself.

Hormones of Pregnancy

Hormones that maintain pregnancy are Human chorionic gonadotropin (HCG), estrogen, progesterone, and human chorionic somatomammotropin (HCS). HCG helps prevent spontaneous abortion by maintaining the corpus luteum which continues to secrete estrogen and progesterone which in turn prevents the release of FSH and LH. HCS from the placenta mobilizes fatty acids as fuel for the mother while it spares glucose for use by the fetus. Placental lactogen stimulates breast development. Relaxin relaxes joints and ligaments to facilitate easier delivery. HCG is the hormone detected in pregnancy tests.

Teratogens

During certain periods of the fetal stage, the fetus is very susceptible to specific teratogens which affect certain organs of development. The first two months are very important to organ development. However, during the entire fetal stage, the fetus's central nervous system is susceptible to developmental disruption. Infectious diseases where the pathogenic organism crosses the placenta during this time of organ development can lead to devastating effects such as blindness, hydrocephalus, cerebral palsy, seizures, and profound mental and physical retardation. Other teratogens, viruses, chemicals, and other agents, can cause anatomical deformities in the fetus. Alcohol abuse causes more birth defects than any other drug. Children borne with fetal alcohol syndrome are characterized with a small head, malformed facial features, central nervous system defects, stunted growth and behavioral problems. Cigarette smoking contributes to fetal and infant mortality.

Birthing Process

During pregnancy, false uterine contractions are felt by the mother. The birthing process (parturition) is marked by the onset of true labor contractions. Contractions usually start about thirty minutes apart but become more intense and rapid (every one to three minutes). Regular contractions of the uterus prepare the uterus for birth. Labor can be divided into three stages:

- Dilation (first stage): the longest stage lasting anywhere from 8-24 hours for a first time mother to a few minutes for a mother who has given birth previously. It is marked by the widening of the cervical

canal and thinning of the cervix (effacement). The amniotic sac usually ruptures at this time.
- Expulsion (second stage) of the fetus can range from minutes to several hours depending on the number of previous births of the mother.
- The placental stage (third stage) lasts anywhere from 5-30 minutes after delivery. The placenta is expelled due to powerful uterine contractions. About 350 ml of blood is lost at this time.

The following is a link to PBS/Nova website. It describes the miracle of life.

http://www.pbs.org/wgbh/nova/miracle/program.html

GENETICS AND GENOMICS

DNA Code

The total amount of DNA in a cell is its genome. DNA is the blueprint that tells the cell through RNA what proteins to make. DNA is composed of two strands of nucleotides. The nitrogenous bases are the purines (adenine & guanine) and pyrimidines (cytosine & thymine). They pair up as A-T or C-G only. Deoxyribose is the sugar present. DNA forms a double alpha helix (a twisted ladder formation). Sections of DNA are the genes (a certain segment of DNA that contains the necessary code to make a protein or RNA molecule). DNA maintains the genetic code during reproduction and still provides variability. A protein's primary structure, the order and type of amino acids in the chain, determines its shape and function. Phenotype is the expression of a gene.

RNA Code

RNA is composed of single strands of nucleotides. The nitrogenous bases of RNA are the purines (adenine & guanine) and pyrimidines (cytosine and uracil). The sugar is always ribose. There are a number of types of RNA, each with a different function. Messenger RNA (mRNA) is produced from DNA patterns in transcription. The master DNA code is first copied onto mRNA through transcription. Transfer RNA (tRNA) converts the master code on mRNA into a specific amino acid. There are 64 varieties of codons determined by anti-codons (nucleotide triplets) and amino acid binding sites. 61/64 codons represent some type of amino acid; the other types are start or stop codons. Ribosomal RNA (rRNA) forms the major part of the ribosome and participates in protein synthesis. Transcription produces mRNA using the DNA code by complimentary base pairing and occurs within the nucleus. mRNA attaches to ribosome. tRNA anticodons attach to complimentary codons of mRNA amino acids join to produce protein. Translation is the production of a protein from a mRNA strand. All elements needed to synthesize a protein are brought together on the ribosome. Translation occurs in the cytoplasm.

Emerging Role of Genetics

Genetics is the study of trait transmission through DNA passed by the gametocytes from generation to generation. Factors that influence development of individual characteristics are heredity and environment. Environment can cause the expression of genes. For example, if a person has the genes to become a tall person but does not has access to a nutritional diet, those genes fail to express themselves fully and the person doesn't develop the height that his genes code. Genes are part of chromosomes. They encode proteins. DNA makes RNA makes proteins. Genomics considers heredity in terms of genes that interact with one another and the environment. Proteomics considers how proteins interact within the body.

Modes of Inheritance

Genotype is the combination of genes within a cell or organism. Phenotype is the physical expression of the genotype. Humans have 22 pairs of autosomes or homologous chromosomes and one pair of sex chromosomes (XY). One chromosome comes from the mother's egg and the other comes from the father's sperm. Genes are located on the chromosomes. An allele is different forms of a gene found at the same locus (*pl. loci)* of a paired chromosome. In homozygous alleles the genes are identical. Heterozygous alleles have genes that are different.

Dominant and Recessive Inheritance

If a gene is dominant, that characteristic is expressed even when heterozygous genes are present. Dominant genes determine the phenotype. If a gene is recessive, the only way this characteristic can be expressed is if it is homozygous. Otherwise the trait is usually masked. Heterozygous means having a dominant and a recessive gene. Sometimes genes are co-dominant that is, two genes, and share dominance such as in AB blood type. Incomplete dominance occurs when the heterozygous phenotype is intermediate between that of either gene. Neither is dominant over the other.

Matters of Sex

Sex is determined at conception by the male gamete. Traits transmitted on X chromosome are X-linked those on the Y-chromosome are Y-linked. Y-linked traits are only transmitted from fathers to sons. Gender can have some effects on phenotype. Sex-limited trait affects a structure or a function that is present only in females or in males. Sex-influenced inheritance the allele is dominant in one sex but recessive in the other. Difference in expression is due to hormones. Genomic imprinting is the expression of a disorder that is dependent on which parent transmits the disease causing gene.

Mutation Mechanisms

A mutation is a permanent change in the DNA that may be passed along from generation to generation. The wild type gene exhibits normal characteristics. A mutant gene usually causes disease or an unusual trait to be expressed. Causes of mutation can be spontaneous, a random change in DNA that arises from mistakes in DNA replication or induced due to chemical or physical factors. Induced mutations usually result in cell death but can lead to cancer.

Categories of mutations include point mutations (a change the nature of one gene, called a frameshift mutation is brought about by the deletion or insertion of a base pair), a substitution mutation (a wrong base pair is put in place of correct bp producing error in base pairing, thus a change in the codon), an inversion mutation (change in one or two codons whereby adjacent base pairs change position). Mutations can be silent (no change in amino acid), missense with consequences from none to severe, or nonsense which leads to a STOP codon. Large mutations are ones in which whole chromosomes are lost or large genetic sequences are inserted and are usually fatal. What genetic mutations in higher animals, such as humans, done to enhance the species? (For example, the ability to form speech and larger brains leading to civilization?)

Disorder terms

Completely penetrant indicates that everyone who has the gene will express it, whereas is a person exhibits incomplete penetrance; they will only have some symptoms. Variably expressive means symptoms vary in intensity in different people (severity). Pleiotrophy occurs when a single genetic disorder can produce several symptoms. (Marfan syndrome affects elastic connective tissue). Complex traits indicate that one or more genes plus the environment mold multifactorial traits. Examples include height and skin color, heart disease, and cancer.

Chromosome Disorders

Polyploidy means that greater than two sets of chromosomes are produced. This is incompatible with life if in embryo. Aneuploidy refers to cells missing a chromosome or having an extra one. This results from nondisjuncture in meiosis. Monosomy in humans is 45 chromosomes (Turner's syndrome XO). Trisomy in humans is 47 chromosomes. Examples include Down's syndrome (Trisomy 21), Trisomy 18, Trisomy 13, and Klinefelter's syndrome XXY.

Prenatal tests to check for possible genetic problems are amniocentesis, chorionic villus sampling, and fetal cell sorting. All carry somewhat of a risk to the fetus.

Gene Therapy can be heritable gene therapy or non-heritable gene therapy.

Watch sequence 1, 2, 3, 6, 9, 10, 11, 12, 13, 14, 15, and 16 from the following website.

http://www.pbs.org/wgbh/nova/genome/program.html

Look at the following website. Go through all the topics. I will think up some wonderful questions from these websites to put on the exam.

http://www.dnaftb.org/dnaftb/1/concept/index.html