LEARNING OBJECTIVES

- Explain the key concepts of basic human genetics.
- Discuss the purpose, key findings, and potential outcomes of the Human Genome Project.
- Describe expanded roles for nurses in genetics and genetic counseling.
- Examine ethical dimensions of genetic screening.
- Summarize the current status of gene therapy (gene transfer).
- Summarize the process of fertilization.
- Discuss the development, structure, and functions of the placenta.
- Describe the composition and functions of the amniotic fluid.
- Identify three organs or tissues arising from each of the three primary germ layers.
- Summarize the significant changes in growth and development of the embryo and fetus.
- Identify the potential effects of teratogens during vulnerable periods of embryonic and fetal development.

KEY TERMS AND DEFINITIONS

- blastocyst: Stage in development of a mammalian embryo, occurring after the morula stage, that consists of an outer layer, or trophoblast, and a hollow sphere of cells enclosing a cavity
- chorionic villi: Tiny vascular protrusions on the chorionic surface that project into the maternal blood sinuses of the uterus and that help form the placenta and secrete human chorionic gonadotropin
- chromosomes: Elements within the cell nucleus carrying genes and composed of DNA and proteins
- conception: Union of the sperm and ovum resulting in fertilization; formation of the one-celled zygote
- decidua basalis: Maternal aspect of the placenta made up of uterine blood vessels, endometrial stroma, and glands; shed in lochial discharge after birth
- embryo: Conceptus from the second or third week of development until approximately the eighth week after conception
- fertilization: Union of an ovum and a sperm
- fetal membranes: Amnion and chorion surrounding the fetus
- fetus: Child in utero from approximately the ninth week after conception until birth
- gamete: Mature male or female germ cell; the mature sperm or ovum
- genetics: Study of single gene or gene sequences and their effects on living organisms
- genome: Complete copy of genetic material in an organism
- genomics: Study of the entire deoxyribonucleic acid (DNA) structure of all of an organism’s genes including functions and interactions of genes
- implantation: Embedding of the fertilized ovum in the uterine mucosa; nidation
- karyotype: Schematic arrangements of the chromosomes within a cell to demonstrate their numbers and morphology
- meiosis: Process by which germ cells divide and decrease their chromosomal numbers by one half
- mitosis: Process of somatic cell division in which a single cell divides, but both of the new cells have the same number of chromosomes as the first
- monosomy: Chromosomal aberration characterized by the absence of one chromosome from the normal diploid complement
- morula: Developmental stage of the fertilized ovum in which there is a solid mass of cells resembling a mulberry
- mosaicism: Condition in which some somatic cells are normal, whereas others show chromosomal aberrations
- sex chromosomes: Chromosomes associated with determination of sex: the X (female) and Y (male) chromosomes; the normal female has two X chromosomes, and the normal male has one X and one Y chromosome
- teratogens: Environmental substances or exposures that result in functional or structural disability
- zygote: Cell formed by the union of two reproductive cells or gametes; the fertilized ovum resulting from the union of a sperm and an ovum
This chapter presents a brief discussion of genetics and the role of the nurse in genetics. It also provides an overview of the process of fertilization and of the development of the normal embryo and fetus.

Genetics is currently recognized as a contributing factor in virtually all human illnesses. In maternity care, genetics issues occur before, during, and after pregnancy (Hamilton & Wynshaw-Boris, 2004). With growing public interest in genetics, increasing commercial pressures, and Web-based opportunities for individuals, families, and communities to participate in the direction and design of their genetic health care, genetic services are rapidly becoming an integral part of routine health care (see Resources at end of this chapter).

For most genetic conditions, therapeutic or preventive measures do not exist or are very limited. Consequently, the most useful means of reducing the incidence of these disorders is by preventing their transmission. It is standard practice to assess all pregnant women for heritable disorders to identify potential problems.

Genetic disease affects people of all ages, from all socioeconomic levels, and from all racial and ethnic backgrounds. Genetic disease affects not only individuals, but also families, communities, and society. Advances in genetic testing and genetically based treatments have altered the care provided to affected individuals. Improvements in diagnostic capability have resulted in earlier diagnosis and enabled individuals who previously would have died in childhood to survive into adulthood. The genetic aberrations that lead to disease are present at birth but may not be manifested for many years, or possibly never manifested.

Some disorders appear more often in ethnic groups. Examples include Tay-Sachs disease in Ashkenazi Jews, Chinese and Thais; neural tube defects in Irish, Scots, and Welsh; phenylketonuria (PKU) in Irish, Scots, Scandinavians, Irish, and Polish; cystic fibrosis (CF) in Caucasians, Ashkenazi Jews, and Hispanics; and Niemann-Pick disease, type A, in Ashkenazi Jews (Hamilton & Wynshaw-Boris, 2004; Jenkins & Wapner, 2004).

Genomics

Genomics address the functions and interactions of all the genes in an organism. It is the study of the entire DNA structure. New fields incorporating genomic knowledge are emerging, for example, nutrigenomics and pharmacogenomics (Horner, 2004). Genomic health care incorporates assessment, diagnosis, and treatment that use information about gene function. It is highly individualized because treatment options are based on the phenotypic responses of an individual. Genetic information includes personal data as well as information about blood relatives (Horner, 2004).

Relevance of Genetics to Nursing

Genetic disorders span every clinical practice specialty and site, including school, clinic, office, hospital, mental health agency, and community health settings. Because the potential impact on families and the community is significant (Box 7-1), genetics must be integrated into nursing education and practice. A genetic paradigm must be embraced; that is, genetic information, technology, and testing must be incorporated into health care services.

Although many of the roles for nurses in genetics are being expanded or developed, all nurses should be prepared to collaborate in interdisciplinary clinical partnerships and provide five main genetics-related nursing activities (International Society of Nurses in Genetics [ISONG], 1998; Lea, Peatham, & Monsen, 2002). The five main activities are as follows:

- Collecting, reporting, and recording genetics information
- Offering genetics information and resources to patients and families
- Participating in the informed consent process and facilitating informed decision making
- Participating in management of patients and families affected by genetic conditions
Potential Impact of Genetic Disease on Family and Community

- Financial cost to family
- Decrease in planned family size
- Loss of geographic mobility
- Decreased opportunities for siblings
- Loss of family integrity
- Loss of career opportunities and job flexibility
- Social isolation
- Lifestyle alterations
- Reduction in contributions to their community by families
- Disruption of husband-wife or partner relationship
- Threatened family self-concept
- Coping with intolerant public attitudes
- Psychologic effects
- Stresses and uncertainty of treatment
- Physical health problems
- Loss of dreams and aspirations
- Cost to society of institutionalization or home or community care
- Cost to society because of additional problems and needs of other family members
- Cost of long-term care
- Housing and living arrangement changes


Genetic History-Taking and Genetic Counseling Services

It is standard practice in obstetrics to determine whether a heritable disorder exists in a couple or in anyone in either of their families. The goal of screening is to detect or define risk for disease in low-risk populations and identify those for whom diagnostic testing may be appropriate. A nurse can obtain a genetics history using a questionnaire or checklist such as the one in Fig. 7-1.

Genetic counseling that follows may occur in the office, or referral to a geneticist may be necessary. The most efficient counseling services are associated with the larger universities and major medical centers. This is also where support services are available (e.g., biochemistry and cytology laboratories), usually from a group of specialists under the leadership of a physician trained in medical genetics. Health professionals should become familiar with people who provide genetic counseling and the places that offer counseling services in their area of practice. See Resources at the end of this chapter for information on genetics resources.

Ethical Considerations

Researchers have proposed using fetal neurologic, liver, and pancreatic tissues to treat adults with Parkinson disease, metabolic disorders, or head and spinal cord injury. The use of fetal tissue in research was banned in the United States for several years, but the ban was lifted in 1993.

Most genetic testing is offered prenatally in order to identify genetic disorders in fetuses (Jenkins & Wapner, 2004). Other requests for genetic testing occur for sex selection or for late-onset disorders. An ethic of social responsibility should guide genetic counselors in their interactions with patients while recognizing that people make their choices by integrating personal values and beliefs with their new knowledge of genetic risk and medical treatments.

Other ethical issues relate to autonomy, privacy, and confidentiality. Should genetic testing be done when there is no treatment available for the disease? When is it appropriate to warn family members at risk for inherited diseases? When should presymptomatic testing be done? Some who might benefit from genetic testing choose not to have it, fearing discrimination based on the risk of a genetic disorder. Several states have prohibitions against insurance discrimination; other states are expected to follow their lead. Until guidelines for genetic testing are created, caution should be exercised. The benefits of testing should be weighed carefully.

Probably the most important of all nursing functions is providing emotional support to the family during all aspects of the counseling process. Feelings that are generated under the real or imagined threat posed by a genetic disorder are as varied as the people being counseled. Responses may include a variety of stress reactions such as apathy, denial, anger, hostility, fear, embarrassment, grief, and loss of self-esteem.
UNIT THREE

PREGNANCY

Risk Factors for Genetic Disorders

Review the following list of risk factors and place a check next to the “yes” responses.

_____ Will you be age 35 or older when your baby is due?

_____ If you or your partner are of Mediterranean or Asian descent, do either of you or anyone in your families have thalassemia?

_____ Is there a family history of neural tube defects?

_____ Have you ever had a child with a neural tube defect?

_____ Is there a family history of congenital heart defects?

_____ Is there a family history of Down syndrome?

_____ Have you ever had a child with Down syndrome?

_____ If you or your partner are of Eastern European Jewish or French Canadian descent, is there a family history of Tay-Sachs?

_____ If you or your partner are of Eastern European Jewish descent, is there a family history of Canavan disease?

_____ If you or your partner are African-American, is there a family history of sickle cell disease or trait?

_____ Is there a family history of hemophilia?

_____ Is there a family history of muscular dystrophy?

_____ Is there a family history of cystic fibrosis?

_____ Is there a family history of Huntington disease?

_____ Is anyone in your or your partner’s family mentally retarded?

_____ If so, was that person tested for Fragile X syndrome?

_____ Do you, your partner, anyone in your families, or any of your children have any other genetic diseases, chromosomal disorders, or birth defects?

_____ Do you have a metabolic disorder such as diabetes or phenylketonuria?

_____ Have you had more than two miscarriages in a row?

_____ Have you ever had a baby who was stillborn?

Fig. 7-1 Questionnaire for identifying couples having increased risk for offspring with genetic disorders.

against the potential for harm. The American Academy of Pediatrics (2001) recommends that children not have genetic testing for disorders that have a late-onset and for which there is no treatment.

Preimplantation genetic diagnosis (PGD) is available in a limited number of centers. In this procedure, embryos are tested before implantation by in vitro fertilization (IVF) (Jones & Fallon, 2002). PGD has the potential to eliminate specific disorders in pregnancies conceived by IVF.

The Human Genome Project

The Human Genome Project began in 1990 as an international effort to map and sequence the genetic makeup of humans; it is funded by the National Institutes of Health (NIH) and the Department of Energy. Initial sequencing of the human genome (the copy of genetic material) was completed in June 2000, well ahead of schedule. A substantially complete version of the human genome was announced in April 2002. The map will facilitate study of hereditary diseases and
will provide the potential for making changes at the gene level to treat or prevent hereditary diseases.

Two key findings from initial efforts to sequence and analyze the human genome are that (1) all human beings are 99.9% identical at the DNA level, and (2) approximately 30,000 to 40,000 genes (pieces or sequences of DNA that contain information needed to make proteins) make up the human genome (International Human Genome Sequencing Consortium, 2001). The finding that human beings are 99.9% identical at the DNA level should help to discourage the use of science as a justification for drawing precise racial boundaries around certain groups of people (Collins & Mansoura, 2001). The vast majority of the 0.1% genetic variations are found within and not among populations. The finding that humans have 30,000 to 40,000 genes, which is only twice as many as roundworms (18,000) and flies (13,000), was unexpected. Scientists had estimated that there were 80,000 to 150,000 genes in the human genome. It had been assumed that the main reason that humans are more evolved and more highly sophisticated than other species is that they have more genes.

Initial efforts to sequence and analyze the human genome have proven invaluable in the identification of genes involved in disease and in the development of genetic tests. More than 100 genes involved in diseases such as Huntington disease (HD), breast cancer, colon cancer, Alzheimer disease, achondroplasia, and cystic fibrosis have been identified. Genetic tests for more than 1446 inherited conditions are commercially available (see Chapter 21); of these, 836 are clinical tests and 110 are research tests (Genetic Tests website, www.genetics.org [accessed July 1, 2005]).

### Genetic testing

Most of the genetic tests now being offered in clinical practice are tests for single-gene disorders in patients with clinical symptoms or who have a family history of a genetic disease (Yoon et al., 2001). Some of these genetic tests are prenatal tests or tests used to identify the genetic status of a pregnancy at risk for a genetic condition. Current prenatal testing options include maternal serum screening (a blood test used to see if a pregnant woman is at increased risk for carrying a fetus with a neural tube defect or a chromosomal abnormality such as Down syndrome) and invasive procedures (amniocentesis and chorionic villus sampling). Other tests are carrier screening tests, which are used to identify individuals who have a gene mutation for a genetic condition but do not show symptoms of the condition because it is a condition that is inherited in an autosomal recessive form (e.g., CF, sickle cell disease, and Tay-Sachs disease). Another type of genetic testing is predictive testing, which is used to clarify the genetic status of asymptomatic family members. The two types of predictive testing are presymptomatic and predispositional. Mutation analysis for HD, a neurodegenerative disorder, is an example of presymptomatic testing. If the gene mutation for HD is present, symptoms of HD are certain to appear if the individual lives long enough. Testing for a BRCA-1 gene mutation to determine breast cancer susceptibility is an example of predispositional testing. Predispositional testing differs from presymptomatic testing in that a positive result (indicating that a BRCA-1 mutation is present) does not indicate a 100% risk of developing the condition (breast cancer).

### Pharmacogenomics

One of the most immediate clinical applications of the Human Genome Project may be pharmacogenomics, or the use of genetic information to individualize drug therapy (Phillips, Venotra, Oren, Lee, & Sades, 2001). There has been speculation that pharmacogenomics may become part of standard practice for a large number of disorders and drugs by 2020 (Collins & McKusick, 2001). The expectation is that by identifying common variants in genes that are associated with the likelihood of a good or bad response to a specific drug, drug prescriptions can be individualized based on the individual’s unique genetic makeup (Roses, 2000). A primary benefit of pharmacogenomics is the potential to reduce adverse drug reactions.

### Gene therapy (gene transfer)

In the early 1990s, a great deal of optimism was felt about the possibility of using genetic information to provide quick solutions to a long list of health problems (Collins & McKusick, 2001). However, the field of gene therapy, also known as gene transfer, has sustained a number of major disappointments during the past few years. Although the early optimism about gene therapy was probably never fully justified, it is likely that the development of safer and more effective methods for gene delivery will ensure a significant role for gene therapy in the treatment of some diseases (Collins & McKusick, 2001). Major challenges include targeting the right gene to the right location in the right cells, expressing the transferred gene at the right time, and minimizing adverse reactions (Brouwer, 2001). Some reports detail exciting possibilities regarding the application of gene therapy for homophilia B (Kay et al., 2000) and severe combined immunodeficiency (Anderson, 2000; Cavazzana-Calvo et al., 2000). According to a scientist who is very active in the field of gene therapy, “Gene therapy will succeed with time. And it is important that it does, because no other area of medicine holds as much promise for providing cures for the many devastating diseases that ravage humankind” (Anderson, 2000).

### Ethical, legal, and social implications

An integral part of the Human Genome Project is the Ethical, Legal, and Social Implications (ELSI) program; 5% of the Human Genome Project budget was designated for the study of ELSI of human genome research. This program addresses the potential that genetic information may be used to discriminate against individuals or for eugenic purposes. Continued awareness of and vigilance against such misuse of information is the collective responsibility of health care providers, ethicists, and society.
Management of Genetic Disorders

At this time, no cures exist for genetic disorders, although remedies can be implemented to prevent or reduce the harmful effects of a few disorders. Structural defects can sometimes be modified to produce normal or near-normal function. Surgical therapy is employed for congenital heart defects and cosmetic defects such as cleft lip. Advances in fetal surgery are occurring. Other conditions are treated with product replacement (e.g., thyroid for hereditary cretinism), diet modification (e.g., low-phenylalanine diet for PKU), and corrective devices for missing limbs. Research is being conducted on methods to influence or change genes directly by placing substitute DNA in the cells of those with a genetic mutation, thereby preventing or curing the disease process or relieving symptoms.

The possibility exists that understanding embryonic stem cells (primitive cells that can develop into all types of body tissue, including muscles, nerves, and bones) will lead to new medical discoveries (Box 7-2). The successful cloning of sheep, cattle, mice, and pigs; the production of rhesus monkeys through nuclear transfer of embryonic cells; and the isolation of stem cells constitute breakthroughs in technology. They also raise other ethical questions. On August 25, 2000, the NIH published guidelines for research using human stem cells (NIH, 2000). The nurse involved in genetics must keep abreast of new developments and be prepared to discuss ethical implications with patients and other health care providers.

BOX 7-2

Stem Cells

Stem cells are able to divide for indefinite periods and can differentiate into the many different types of cells that make up an organism. Embryonic stem cells are derived from the blastocyst before it implants in the uterine wall. A zygote is described as totipotent because it has the potential to produce all the cells and tissues that compose an embryo and to support its in utero development. The term pluripotent is used to describe stem cells that generate cells derived from the three embryonic germ layers (endoderm, mesoderm, and ectoderm). The embryonic stem cell is pluripotent. Human stem cells were derived and maintained for the first time in 1998 by Thomson and colleagues by using blastocysts donated by couples undergoing in vitro fertilization. Potential uses of human embryonic stem cells include transplant therapy, in which tissues damaged by disease or injury (e.g., as in diabetes, Parkinson disease, heart disease, and multiple sclerosis) are replaced or restored. Stem cell research engenders ethical concerns related to the source of human embryonic stem cells (embryos left over from in vitro fertilization and aborted fetuses). Currently federal support for research is restricted to existing cell lines.

Estimation of risk

The risks of recurrence of a genetic disorder are determined by the mode of inheritance. The risk of recurrence for disorders caused by a factor that segregates during cell division (i.e., genes and chromosomes) can be estimated with a high degree of accuracy by application of mendelian principles. In a dominant disorder the risk is 50%, or one in two, that a subsequent offspring will be affected; an autosomal recessive disease carries a one-in-four risk of recurrence; and an X-linked disorder is related to the child’s sex, as described in the section related to X-linked inheritance. Translocation chromosomes have a high risk of recurrence.

Disorders in which a subsequent pregnancy would carry no more risk than there is for pregnancy alone (estimated at 1 in 30) include those resulting from isolated incidences not likely to be present in another pregnancy. These disorders include maternal infections (e.g., rubella and toxoplasmosis), maternal ingestion of drugs, most chromosomal abnormalities, and a disorder determined to be the result of a fresh mutation.

Interpretation of risk

Counselors explain the risk estimates to patients without making recommendations or decisions and without allowing their own biases to interfere. The counselor provides appropriate information about the nature of the disorder, the extent of the risks in the specific case, the probable consequences, and (if appropriate) alternative options available; however, the final decision to become pregnant or to continue a pregnancy must be left to the family. An important nursing role is reinforcing the information the families are given and continuing to interpret this information at their level of understanding.

The most important concept that must be emphasized to families is that each pregnancy is an independent event. For example, in monogenic disorders, in which the risk factor is one in four that the child will be affected, the risk remains the same no matter how many affected children are already in the family. Families may make the erroneous assumption that the presence of one affected child ensures that the next three will be free of the disorder. However, “chance has no memory.” The risk is one in four for each subsequent pregnancy. On the other hand, in a family with a child who has a disorder with multifactorial causes, the risk increases with each subsequent child born with the disorder.

Genes and Chromosomes

The hereditary material carried in the nucleus of each somatic (body) cell determines an individual’s physical characteristics. This material—DNA—forms threadlike strands known as chromosomes. Each chromosome is composed of many smaller segments of DNA referred to as genes. Genes or combinations of genes contain coded information that determines an individual’s unique characteristics. The code consists of the specific linear order of the molecules that combine to form the strands of DNA. Genes never act in
isolation; they always interact with other genes and the environment.

All normal human somatic cells contain 46 chromosomes arranged as 23 pairs of homologous (matched) chromosomes; one chromosome of each pair is inherited from each parent. There are 22 pairs of autosomes, which control most traits in the body, and one pair of sex chromosomes, which determines sex and some other traits. The large female chromosome is called the X; the tiny male chromosome is the Y. When one X chromosome and one Y chromosome are present, the embryo develops as a male. When two X chromosomes are present, the embryo develops as a female.

Because each gene occupies a specific chromosome location, and because chromosomes are inherited as homologous pairs, each person has two genes for every trait. In other words, if an autosome has a gene for hair color, its partner also has a gene for hair color at the same location on the chromosome. Although both genes code for hair color, they may not code for the same hair color. Different genes coding for different variations of the same trait are called alleles. An individual with two copies of the same allele for a given trait is said to be homozygous for that trait. With two different alleles, the person is said to be heterozygous for the trait.

The term genotype typically is used to refer to the genetic makeup of an individual when discussing a specific gene pair, but at times, genotype is used to refer to an individual’s entire genetic makeup or all the genes that the individual can pass on to future generations. Phenotype refers to the observable expression of an individual’s genotype, such as physical features, a biochemical or molecular trait, and even a psychological trait. A trait or disorder is considered dominant if it is expressed or phenotypically apparent when only one copy of the gene is present. It is considered recessive if it is expressed only when two copies of the gene are present.

The pictorial analysis of the number, form, and size of an individual’s chromosomes is known as a karyotype. Cells from any nucleated, replicating body tissue (not red blood cells, nerves, or muscles) can be used (Scheuerle, 2001). The most commonly used tissues are white blood cells and fetal cells in amniotic fluid. The cells are grown in a culture and arrested when they are in metaphase, and then the cells are dropped onto a slide. This breaks the cell membranes and spreads the chromosomes, making them easier to visualize. The cells are stained with special stains (e.g., Giemsa stain) that create striping or “banding” patterns. Once the chromosome spreads are photographed or scanned by a computer, they are cut out and arranged in a specific numeric order according to their length and shape. The chromosomes are numbered from largest to smallest, 1 to 22, and the sex chromosomes are designated by the letter X or Y. Each chromosome is divided into two “arms” designated by p (short arm) and q (long arm). A female karyotype is designated as 46,XX and a male karyotype is designated as 46,XY. Fig. 7.2 illustrates the chromosomes in a body cell and a karyotype. Karyotypes can be used to determine the sex of a child and the presence of any gross chromosomal abnormalities.

**Chromosomal Abnormalities**

Chromosomal abnormalities occur in 0.5% to 0.6% of newborn infants; most of these have no significant physical abnormality associated with the defect. The incidence of

![Fig. 7-2 Chromosomes during cell division. A, Example of photomicrograph. B, Chromosomes arranged in karyotype; female and male sex-determining chromosomes.](image-url)
Abnormalities of chromosome number. 

Aneuploidy denotes the correct number of chromosomes. Deviations from the correct number of chromosomes can be one of two types: (1) polyploidy, in which the deviation is an exact multiple of the haploid number of chromosomes; or (2) aneuploidy, in which the numerical deviation is not an exact multiple of the haploid set (Hamilton & Wynshaw-Boris, 2004). Aneuploidy is the most commonly identified chromosome abnormality in humans. Aneuploidy occurs in at least 5% of all clinically recognized pregnancies, and it is the leading known cause of pregnancy loss (Hassold & Hunt, 2001). Aneuploidy also is the leading genetic cause of mental retardation. The two most common aneuploid conditions are monosomies and trisomies. A monosomy is the product of the union between a normal gamete and a gamete that is missing a chromosome. Monosomic individuals only have 45 chromosomes in each of their cells. Limited data are available concerning the origin of monosomies because when an embryo is missing an autosomal chromosome, the embryo never survives. The product of the union of a normal gamete with a gamete containing an extra chromosome is a trisomy. Trisomies are more common than monosomies. Trisomic individuals have 47 chromosomes in each of their cells. Most trisomies are caused by nondisjunction during the first meiotic division. That is, one pair of chromosomes fails to separate. One of the resulting cells contains two chromosomes, and the other contains none. The most common trisomol abnormality is Down syndrome, or trisomy 21 (see discussion in Chapter 27; Nursing Plan of Care). Other autosomal trisomies that have been identified are trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome). Both conditions have a very poor prognosis, and most affected children die from cardiac or respiratory complications within 6 months of birth. Nondisjunction can also occur during mitosis. If this occurs early in development, when cell lines are forming, the individual has a mixture of cells, some with a normal number of chromosomes and others either missing a chromosome or containing an extra chromosome. This condition is known as mosaicism.

Sex chromosome abnormalities

Sex chromosome abnormalities are caused by nondisjunction during gametogenesis in either parent. The most common deviation in females is Turner syndrome, or monosomy X (having only one X chromosome); the affected female exhibits juvenile external genitalia with undeveloped ovaries. She is usually short in stature with webbing of the neck. Intelligence may be impaired. Most affected embryos miscarry spontaneously. The most common deviation in males is Klinefelter syndrome, or trisomy XXX. The affected male has poorly developed secondary sexual characteristics and small testes. He is infertile, usually tall, and effeminate. Males who are mosaic for Klinefelter syndrome may be fertile. Subnormal intelligence is usually present.

Patterns of Genetic Transmission

Heritable characteristics are those that can be passed on to offspring. The patterns by which genetic material is transmitted to the next generation are affected by the number of genes involved in the expression of the trait. Many phenotypic characteristics result from two or more genes on different chromosomes acting together (referred to as multifactorial inheritance); others are controlled by a single gene (unifactorial inheritance). Defects at the gene level cannot be determined by conventional laboratory methods such as karyotyping. Instead, genetic specialists predict the probability of the presence of an abnormal gene from the known occurrence of the trait in the individual’s family and the known patterns by which the trait is inherited.
Multifactorial inheritance
Most common congenital malformations, such as cleft lip and palate and neural tube defects, result from multifactorial inheritance, a combination of genetic and environmental factors. Each malformation may range from mild to severe, depending on the number of genes for the defect present or the amount of environmental influence. Multifactorial disorders tend to occur in families. Some malformations occur more often in one sex than the other. For example, pyloric stenosis and cleft lip are more common in males, and cleft palate is more common in females.

Unifactorial inheritance
If a single gene controls a particular trait, disorder, or defect, its pattern of inheritance is referred to as unifactorial mendelian or single-gene inheritance. The number of unifactorial abnormalities far exceeds the number of chromosomal abnormalities. This is understandable, considering that 30,000 to 40,000 genes in the haploid number (23) of chromosomes are passed on to an offspring from each parent.

Unifactorial or single-gene disorders follow the inheritance patterns of dominance, segregation, and independent assortment described by Mendel and include autosomal dominant, autosomal recessive, and X-linked dominant and recessive modes of inheritance (Fig. 7-3).

Autosomal dominant inheritance. Autosomal dominant inheritance disorders are those in which the abnormal gene for the trait is expressed even when the other member of the pair is normal. The abnormal gene may appear as a result of a mutation, a spontaneous and permanent change in the normal gene structure. In this case the disorder occurs for the first time in the family. Usually an affected individual comes from multiple generations having the disorder (see Fig. 7-3, B and C). Males and females are equally affected.

Examples of common autosomal dominantly inherited disorders are Marfan syndrome (a disorder of connective tissue resulting in skeletal, ocular, and cardiovascular abnormalities), achondroplasia (dwarfism), polydactyly (extra digits), Huntington disease, and polycystic kidney disease.

Neurofibromatosis (NF) is a progressive disorder of the nervous system that causes tumors to form on nerves anywhere in the body. NF affects all races, all ethnic groups, and both sexes equally. Half of the cases of NF result from spontaneous genetic mutation, whereas the other half are inherited in an autosomal dominant manner. Two genetically distinct forms of NF are NF1, the most common type, with an incidence of 1 in 3000 (Mueller & Young, 2001), and NF2, with an incidence of 1 in 35,000. The most notable features of NF1 are the small pigmented skin lesions known as café-au-lait spots and the neurofibromata (small, soft, fleshy growths). Individuals with NF1 generally are able to live a normal, healthy life. Café-au-lait spots and neurofibromata can occur with NF2, but they are far less common than with NF1.

Autosomal recessive inheritance. Autosomal recessive inheritance disorders are those in which both genes of a pair must be abnormal for the disorder to be expressed. Heterozygous individuals have only one abnormal gene and are unaffected clinically because their normal gene overshadows the abnormal gene. They are known as carriers of the recessive trait. For the trait to be expressed, two carriers must each contribute the abnormal gene to the offspring (Fig. 7-3, C). Males and females are equally affected. Most inborn errors of metabolism, such as PKU, galactosemia, maple syrup urine disease, Tay-Sachs disease, sickle cell anemia, and CF, are autosomal recessive inherited disorders.

X-linked dominant inheritance. X-linked dominant inheritance disorders occur in males and heterozygous females. Because the females also have a normal gene, the effects are less severe than in affected males. Affected males transmit the abnormal gene only to their daughters, on the X chromosome. Fragile X syndrome and vitamin D-resistant rickets are examples of X-linked dominant inherited disorders.

X-linked recessive inheritance. Abnormal genes for X-linked recessive inheritance disorders are carried on the X chromosome. Females may be heterozygous or homozygous for traits carried on the X chromosome because they have two X chromosomes. Males are hemizygous because they have only one X chromosome carrying genes, with no alleles on the Y chromosome. Therefore X-linked recessive disorders are most often manifested in the male with the abnormal gene on his single X chromosome. Hemophilia, color blindness, and Duchenne muscular dystrophy are all X-linked recessive disorders.

Fig. 7-3 Possible offspring in three types of matings. A, Homozygous-dominant parent and homozygous-recessive parent. Children: all heterozygous, displaying dominant trait. B, Heterozygous parent and homozygous-recessive parent. Children: 50% heterozygous, displaying dominant trait; 50% homozygous, displaying recessive trait. C, Both parents heterozygous. Children: 25% homozygous, displaying dominant trait; 25% homozygous, displaying recessive trait; 50% heterozygous, displaying dominant trait.
EVIDENCE-BASED PRACTICE

Folate Supplements to Prevent Neural Tube Defects

BACKGROUND
- Within 3 weeks of implantation, the embryonic neural groove is formed along the dorsum and will give rise to the brain and spinal cord. The groove folds into a tube by 1 month postfertilization (6 weeks from last menstrual period [LMP]). Defects in this closure can lead to anencephaly (absence of brain, cranial vault, and covering skin), which is incompatible with survival, and spina bifida (herniation of the spinal cord and/or meninges), which has a high mortality rate and mild to extreme neural damage. Neural tube defects (NTDs) may be attributed to both genetic and environmental causes. Screening during the second trimester, using maternal serum alpha-fetoprotein estimation or fetal ultrasound, can identify NTDs while the pregnancy can still be terminated. This has led to a decrease in birth rates of infants with NTDs, but it does not address primary prevention.

OBJECTIVES
- The reviewers’ primary objective was to inquire as to whether periconceptional folate or multivitamins can decrease the prevalence of NTDs.
- Interventions were the folate supplements or multivitamins. Outcomes, aside from NTDs, might include other defects, miscarriage, multiple pregnancy, preterm birth, perinatal and infant mortality, length of supplementation before conception, blood and tissue levels of folate, and attitudes toward and knowledge of NTDs in the population and among practitioners.

METHODS
Search Strategy
- The reviewers searched Cochrane, MEDLINE, and Zetoc, using the search keywords “neural tube defects.”
- Four randomized or quasi-randomized, controlled trials, representing 6,425 women, were selected. One dissemination trial looked at the impact of printed materials on six communities. The trials took place in Hungary, the Republic of Ireland, the United Kingdom, Israel, Australia, Canada, the former USSR, and France, from 1981 to 1994.

Statistical Analyses
- Statistical analyses of homogeneous data were performed, using relative risks with 95% confidence intervals.

FINDINGS
- NTDs were significantly decreased by periconceptional supplementation. There was a consistent increase in the incidence of NTDs.
- Higher dietary folate, taken periconceptionally (before conception and during the first 2 months of pregnancy), seems to greatly decrease the incidence of NTDs.

CONCLUSIONS
- There is evidence that folate supplementation reduces the incidence of NTDs.

IMPLICATIONS FOR PRACTICE
- Most national policy statements call for 0.4 mg/day for all childbearing-age women, especially if they are contemplating pregnancy. For women with a history of an affected prior pregnancy, the recommended dose is 4 mg/day. Many women are not aware that folate was required periconceptionally for effective prevention of NTDs.
- Randomization was not clear, nor was the dropout rate discussed in the review. These studies did not reach those whose pregnancy was unplanned, which can number half of all pregnancies in some areas. There was little information about the effects of folate on other drugs. None of the trials looked at dietary folate, which may be more practical in certain developing countries than supplementation.

LIMITATIONS
- Multivitamins alone did not decrease the rate of NTDs.
- There is evidence that folate supplementation reduces the incidence of NTDs.
- Higher dietary folate, taken periconceptionally (before conception and during the first 2 months of pregnancy), seems to greatly decrease the incidence of NTDs.

IMPLICATIONS FOR FURTHER RESEARCH
- New trials on cost-effective information dissemination can identify the most effective methods to educate childbearing-age women so that they can take folate before pregnancy. Large randomized, controlled trials can determine any side effects of folate, such as multiple births, which could change the relative risks and benefits of folate supplementation substantially. Trials involving food products that contain added folate would be informative. Folate deficiency recently has been tied to cardiovascular disease. Some trials in the future may establish the need for increased folate throughout life.

Inborn errors of metabolism

Disorders of protein, fat, or carbohydrate metabolism that reflect absent or defective enzymes generally follow a recessive pattern of inheritance. Enzymes, the actions of which are genetically determined, are essential for all the physical and chemical processes that sustain body systems. Defective enzyme action interrupts the normal series of chemical reactions from the affected point onward. The result may be an accumulation of...
a damaging product such as phenylalanine or the absence of a necessary product such as thyroxin or melanin.

Phenylketonuria (PKU) is an uncommon disorder caused by autosomal recessive genes. A deficiency in the liver enzyme phenylalanine hydroxylase results in failure to metabolize the amino acid phenylalanine, allowing its metabolites to accumulate in the blood. The incidence of this disorder is 1 in every 10,000 to 20,000 births. The highest incidence is found in Caucasians (from northern Europe and the United States). It is rarely seen in Jewish, African, or Japanese populations. Screening for PKU is routinely performed on all infants through a blood test.

Tay-Sachs disease, inherited as an autosomal recessive trait, results from a deficiency in hexosaminidase. It occurs
more commonly in Ashkenazi Jews and French-Canadians from Quebec. Infants appear normal until 4 to 6 months of age, then the clinical symptoms appear: apathy and regression in motor and social development, and decreased vision. Death occurs between ages 3 and 4 years. No treatment exists.

CF (mucoviscidosis or fibrocystic disease of the pancreas) is inherited as an autosomal recessive trait and is characterized by generalized involvement of exocrine glands. Clinical features are related to the altered viscosity of mucus-secreting glands throughout the body. Overall incidence is 1 per every 2000 births. Advances in diagnosis and treatment have improved the prognosis; many affected individuals live to adulthood. Some affected women have borne children, but men generally are sterile.

Meconium ileus occurs in about 10% of newborns with CF. Although an initial stool may be passed from the rectum with none thereafter, usually no meconium is passed during the first 24 to 48 hours. The abdomen becomes increasingly distended, and eventually the newborn requires a laparotomy for diagnosis and treatment of the condition. (See discussion in Chapter 27.)

**Nongenetic Factors Influencing Development**

Not all congenital disorders are inherited. Congenital means that the condition was present at birth. Some congenital malformations may be the result of teratogens, that is, environmental substances or exposures that result in functional or structural disability. In contrast to other forms of developmental disabilities, disabilities caused by teratogens are, in theory, totally preventable. Known human teratogens are drugs and chemicals, infections, exposure to radiation, and certain maternal conditions such as diabetes and PKU (Box 7-3). A teratogen has the greatest effect on the organs and parts of an embryo during its periods of rapid differentiation. This occurs during the embryonic period, specifically from days 15 to 60. Brain growth and development continue during the fetal period, and teratogens can severely affect mental development throughout gestation (Fig. 7-4).

In addition to genetic makeup and the influence of teratogens, the adequacy of maternal nutrition influences development. The embryo and fetus must obtain the nutrients they need from the mother’s diet; they cannot tap the maternal reserves. Malnutrition during pregnancy produces low-birth-weight newborns who are susceptible to infection. Malnutrition also affects brain development during the latter half of gestation and may result in learning disabilities in the child. Inadequate folic acid is associated with neural tube defects.

The field of human behavioral genetics seeks to understand genetic and environmental influences on variations in human behavior (McInerney, 2004). Behavior involves multiple genes. Study of behavior and genes requires analysis of families and populations to compare those who have the trait with those who do not. The result is an estimate of the amount of variation in the population attributable to genetic factors. The findings of this research have significant political and social implications. For example, what are the social consequences of determining a genetic diagnosis of traits such as intelligence, criminality, or homosexuality? Caution must be exercised in accepting discoveries in behavioral genetics until there is substantial scientific corroboration (McInerney & Rothstein, 2004).

**Box 7-3**

### Etiology of Human Malformations

<table>
<thead>
<tr>
<th>Etiology</th>
<th>Malformed Live Births (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Environmental</td>
<td>10</td>
</tr>
<tr>
<td>Maternal conditions</td>
<td>4</td>
</tr>
<tr>
<td>Alcoholism, diabetes, endocrinopathies, phenylketonuria, smoking, nutritional problems</td>
<td>4</td>
</tr>
<tr>
<td>Infectious agents</td>
<td>3</td>
</tr>
<tr>
<td>Rubella, toxoplasmosis, syphilis, herpes simplex, cytomegalic inclusion disease, varicella, Venezuelan equine encephalitis</td>
<td>3</td>
</tr>
<tr>
<td>Mechanical problems (deformations)</td>
<td>2</td>
</tr>
<tr>
<td>Anomastic band constrictions, umbilical cord constraint, disparity in uterine size and uterine contents</td>
<td>2</td>
</tr>
<tr>
<td>Chemicals, drugs, radiation, hyperthermia</td>
<td>1</td>
</tr>
<tr>
<td>Genetic</td>
<td>20-25</td>
</tr>
<tr>
<td>Single-gene disorders</td>
<td></td>
</tr>
<tr>
<td>Chromosomal abnormalities</td>
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</tr>
<tr>
<td>Unknown</td>
<td>65-70</td>
</tr>
<tr>
<td>“Spontaneous” errors of development</td>
<td></td>
</tr>
<tr>
<td>Polygenic or multifactorial (gene-environment interactions)</td>
<td></td>
</tr>
<tr>
<td>Other unknowns</td>
<td></td>
</tr>
</tbody>
</table>

Fig. 7-4 Sensitive, or critical, periods in human development. Dark color denotes highly sensitive periods; light color indicates stages that are less sensitive to teratogens. (From Moore, K., & Persaud, T. [2003]. Before we are born: Essentials of embryology and birth defects [6th ed.]. Philadelphia: Saunders.)
CONCEPTION

Cell Division

Cells are reproduced by two different methods: mitosis and meiosis. In **mitosis** the body cells replicate to yield two cells with the same genetic makeup as the parent cell. First the cell makes a copy of its DNA; then it divides, with each daughter cell receiving one copy of the genetic material. **Mitotic division** facilitates growth and development or cell replacement.

**Meiosis**, the process by which germ cells divide and decrease their chromosomal number by half, produces gametes (eggs and sperm). Each homologous pair of chromosomes contains one chromosome received from the mother and one from the father; thus meiosis results in cells that contain one of each of the 23 pairs of chromosomes. Because these germ cells contain 23 single chromosomes, half of the genetic material of a normal somatic cell, they are called **haploid**. When the female gamete (egg or ovum) and the male gamete (spermatozoon) unite to form the zygote, the diploid number of human chromosomes (46, or 23 pairs) is restored. The process of DNA replication and cell division in **meiosis** allows different alleles for genes to be distributed at random by each parent and then rearranged on the paired chromosomes. The chromosomes then separate and proceed to different gametes. Because the two parents have genotypes derived from four different grandparents, many combinations of genes on each chromosome are possible. This random mixing of alleles accounts for the variation of traits seen in the offspring of the same two parents.

Gametogenesis

When a male reaches puberty, his testes begin the process of spermatogenesis. The cells that undergo meiosis in the male are called **spermatocytes**. The primary spermatocyte, which undergoes the first meiotic division, contains the diploid number of chromosomes. The cell has already copied its DNA before division, so four alleles for each gene are

---

**Fig. 7-5** Spermatogenesis. **A**, Gametogenesis in the male produces four mature gametes, the sperm. **B**, Oogenesis. Gametogenesis in the female produces one mature ovum and three polar bodies. Note relative difference in overall size between ovum and sperm. **C**, Fertilization results in the single-cell zygote and restoration of the diploid number of chromosomes.
present. Because the copies are bound together (i.e., one allele plus its copy on each chromosome), the cell is still considered diploid.

During the first meiotic division, two haploid secondary spermatocytes are formed. Each secondary spermatocyte contains 22 autosomes and one sex chromosome; one contains the X chromosome (plus its copy) and the other the Y chromosome (plus its copy). During the second meiotic division the male produces two gametes with an X chromosome and two gametes with a Y chromosome, all of which will develop into viable sperm (Fig. 7-5, A).

Oogenesis, the process of ovum formation, begins during fetal life of the female. All the cells that may undergo meiosis in a woman’s lifetime are contained in her ovaries at birth. The majority of the estimated 2 million primary oocytes (the cells that undergo the first meiotic division) degenerate spontaneously. Only 400 to 500 ova will mature during the approximately 35 years of a woman’s reproductive life. The primary oocytes begin the first meiotic division (i.e., they replicate their DNA) during fetal life but remain suspended at this stage until puberty (Fig. 7-5, B). Then, usually monthly, one primary oocyte matures and completes the first meiotic division, yielding two unequal cells: the secondary oocyte and a small polar body. Both contain 22 autosomes and one X sex chromosome.

At ovulation the second meiotic division begins. However, the ovum does not complete the second meiotic division unless fertilization occurs. At fertilization, a second polar body and the zygote (the united egg and sperm) are produced (Fig. 7-5, C). The three polar bodies degenerate. If fertilization does not occur, the ovum also degenerates.

**Conception**

Conception, defined as the union of a single egg and sperm, marks the beginning of a pregnancy. Conception occurs not as an isolated event but as part of a sequential process. This sequential process includes gamete (egg and sperm) formation, ovulation (release of the egg), union of the gametes (which results in an embryo), and implantation in the uterus.

**Ovum**

Each month, one ovum matures with a host of surrounding supportive cells. At ovulation the ovum is released from the ruptured ovarian follicle. High estrogen levels increase the motility of the uterine tubes so that their cilia are able to capture the ovum and propel it through the tube toward the uterine cavity. An ovum cannot move by itself.

Two protective layers surround the ovum (Fig. 7-6). The inner layer is a thick, acellular layer called the zona pellucida. The outer layer, called the corona radiata, is composed of elongated cells.

Ova are considered fertile for about 24 hours after ovulation. If unfertilized by a sperm, the ovum degenerates and is reabsorbed.

**Sperm**

Ejaculation during sexual intercourse normally propels almost a teaspoon of semen containing as many as 200 to 500 million sperm into the vagina. The sperm swim by means of the flagellar movement of their tails. Some sperm can reach the site of fertilization within 5 minutes, but average transit time is 4 to 6 hours. Sperm remain viable within the woman’s reproductive system for an average of 2 to 3 days. Most sperm are lost in the vagina, within the cervical mucus, or in the endometrium; or they enter the tube that contains no ovum.

As sperm travel through the female reproductive tract, enzymes are produced to aid in their capacitation. Capacitation is a physiologic change that removes the protective coating from the heads of the sperm. Small perforations then form in the acrosome (a cap on the sperm) and allow enzymes (e.g., hyaluronidase) to escape. These enzymes are necessary for the sperm to penetrate the protective layers of the ovum before fertilization.

**Fertilization**

Fertilization takes place in the ampulla (the outer third) of the uterine tube. When a sperm successfully penetrates the membrane surrounding the ovum, both sperm and ovum are enclosed within the membrane, and the membrane becomes impermeable to other sperm; this process is termed the zona reaction. The second meiotic division of the oocyte is then completed, and the ovum nucleus becomes the female pronucleus. The head of the sperm enlarges to become the male pronucleus, and the tail degenerates. The nuclei fuse and the chromosomes combine, restoring the diploid number (46) (Fig. 7-7). Conception, the formation of the zygote (the first cell of the new individual), has been achieved.

Mitotic cellular replication, called cleavage, begins as the zygote travels the length of the uterine tube into the uterus. This voyage takes 3 to 4 days. Because the fertilized egg divides rapidly with no increase in size, successively smaller cells,
called blastomeres, are formed with each division. A 16-cell morula, a solid ball of cells, is produced within 3 days and is still surrounded by the protective zona pellucida (Fig. 7-8, A).

Further development occurs as the morula floats freely within the uterus. Fluid passes through the zona pellucida into the intercellular spaces between the blastomeres, separating them into two parts: the trophoblast (which gives rise to the placenta) and the embryoblast (which gives rise to the embryo). A cavity forms within the cell mass as the spaces come together, forming a structure called the blastocyst cavity. When the cavity becomes recognizable, the whole structure of the developing embryo is known as the blastocyst. Stem cells are derived from the inner cell mass of the blastocyst. The outer layer of cells surrounding the cavity is the trophoblast.

**Implantation**

The zona pellucida degenerates, and the trophoblast attaches itself to the uterine endometrium, usually in the anterior or posterior fundal region. Between 6 and 10 days after conception, the trophoblast secretes enzymes that enable it to burrow into the endometrium until the entire blastocyst is covered. This is known as implantation. Endometrial blood vessels erode, and some women experience slight implantation bleeding (slight spotting and bleeding during the time of the first missed menstrual period). Chorionic villi, or fingerlike projections, develop out of the trophoblast and extend into the blood-filled spaces of the endometrium. These villi are vascular processes that obtain oxygen and nutrients from the maternal bloodstream and dispose of carbon dioxide and waste products into the maternal blood.

After implantation the endometrium is called the decidua. The portion directly under the blastocyst, where the chorionic villi tap into the maternal blood vessels, is the decidua basalis. The portion covering the blastocyst is the decidua capsularis, and the portion lining the rest of the uterus is the decidua vera (Fig. 7-9).
Pregnancy lasts approximately 10 lunar months, 9 calendar months, 40 weeks, or 280 days. Length of pregnancy is computed from the first day of the last menstrual period (LMP) until the day of birth. However, conception occurs approximately 2 weeks after the first day of the LMP. Thus the postconception age of the fetus is 2 weeks less, for a total of 266 days or 38 weeks. Postconception age is used in the discussion of fetal development.

Intrauterine development is divided into three stages: ovum or preembryonic, embryo, and fetus (see Fig. 7-4). The stage of the ovum lasts from conception until day 14. This period covers cellular replication, blastocyst formation, initial development of the embryonic membranes, and establishment of the primary germ layers.

**Primary Germ Layers**

During the third week after conception the embryonic disk differentiates into three primary germ layers: the **ectoderm**, **mesoderm**, and **endoderm** (or entoderm) (see Fig. 7-8, B). All tissues and organs of the embryo develop from these three layers.

The ectoderm, or upper layer of the embryonic disk, gives rise to the epidermis, glands (anterior pituitary, cutaneous, and mammary), nails and hair, central and peripheral nervous systems, lens of the eye, tooth enamel, and floor of the amniotic cavity.

The mesoderm, or middle layer, develops into the bones and teeth, muscles (skeletal, smooth, and cardiac), dermis and connective tissue, cardiovascular system and spleen, and urogenital system.

The endoderm, or lower layer, gives rise to the epithelium lining the respiratory tract and digestive tract, including the oropharynx, liver and pancreas, urethra, bladder, and vagina. The endoderm forms the roof of the yolk sac.

**Development of the Embryo**

The stage of the embryo lasts from day 15 until approximately 8 weeks after conception, when the embryo measures approximately 3 cm from crown to rump. The embryonic stage is the most critical time in the development of the organ systems and the main external features. Developing areas with rapid cell division are the most vulnerable to malformation by environmental teratogens. At the end of the eighth week, all organ systems and external structures are present, and the embryo is unmistakably human (see Fig. 7-4).

**Membranes**

At the time of implantation, two fetal membranes that will surround the developing embryo begin to form. The chorion develops from the trophoblast and contains the chorionic villi on its surface. The villi burrow into the decidua basalis and increase in size and complexity as the vascular processes develop into the placenta. The chorion becomes the covering.
Amniotic Fluid

Amniotic fluid develops as the amnion forms on the side opposite the developing embryonic disk. The developing embryonic disk, amniotic cavity and amnion are formed between this inner cell mass and the outer layer of cells (trophoblast) by the amniotic cavity (see Fig. 7-8, B). As it grows larger, the amnion forms on the side opposite the developing blastocyst (see Fig. 7-8, B, and Fig. 7-9). The developing embryo draws the amnion around itself to form a fluid-filled sac. The amnion becomes the covering of the umbilical cord and covers the chorion on the fetal surface of the placenta. As the embryo grows larger, the amnion enlarges to accommodate the embryo/fetus and the surrounding amniotic fluid. The amnion eventually comes in contact with the chorion surrounding the fetus.

Critical Thinking Exercise

Ultrasound Dating of Pregnancy

Adrienne believes she is 8 weeks pregnant, but her obstetrician believes she is closer to 12 weeks of gestation. Adrienne has come to the clinic for an ultrasound examination for dating. She has many questions for the nurse: How can they tell the length of gestation? What would the fetus look like at this time if she is at 8 weeks of gestation? If she is at 12 weeks of gestation? What fetal structures would be apparent on ultrasound if she is 8 weeks pregnant? If she is 12 weeks pregnant? Would any structural anomalies be apparent at 8 weeks? At 12 weeks? Why is it important to date a pregnancy accurately?

What information should the nurse provide Adrienne?

1. Evidence—Is there sufficient evidence to draw conclusions about what information the nurse should provide Adrienne?
2. Assumptions—What assumptions can be made about the following factors:
   a. Adrienne’s motivation to learn about fetal development
   b. Adrienne’s understanding of fetal development
   c. Adrienne’s knowledge about ultrasound examination?
   d. Why dating the pregnancy is important
3. What implications and priorities for nursing care can be drawn at this time?
4. Does the evidence objectively support your conclusion?
5. Are there alternative perspectives to your conclusion?

of the fetal side of the placenta. It contains the major umbilical blood vessels that branch out over the surface of the placenta. As the embryo grows, the decidua capsularis stretches. The chorionic villi on this side atrophy and degenerate, leaving a smooth chorionic membrane.

The inner cell membrane, the amnion, develops from the interior cells of the blastocyst. The cavity that develops between this inner cell mass and the outer layer of cells (trophoblast) is the amniotic cavity (see Fig. 7-8, B). As it grows larger, the amnion forms on the side opposite the developing blastocyst (see Fig. 7-8, B, and Fig. 7-9). The developing embryo draws the amnion around itself to form a fluid-filled sac. The amnion becomes the covering of the umbilical cord and covers the chorion on the fetal surface of the placenta. As the embryo grows larger, the amnion enlarges to accommodate the embryo/fetus and the surrounding amniotic fluid. The amnion eventually comes in contact with the chorion surrounding the fetus.

Amniotic fluid helps maintain a constant body temperature. It serves as a source of oral fluid and as a repository for waste. It cushions the fetus from trauma by blunting and dispersing outside forces. It allows freedom of movement for musculoskeletal development. The fluid keeps the embryo from tangling with the membranes, facilitating symmetric growth of the fetus. If the embryo does become tangled with the membranes, amputations of extremities or other deformities can occur from constricting amniotic bands.

The volume of amniotic fluid is an important factor in assessing fetal well-being. Having less than 300 ml of amniotic fluid (oligohydramnios) is associated with fetal renal abnormalities. Having more than 2 L of amniotic fluid (hydramnios) is associated with gastrointestinal and other malformations.

Amniotic fluid contains albumin, urea, uric acid, creatinine, lecithin, sphingomyelin, bilirubin, fructose, fat, leukocytes, proteins, epithelial cells, enzymes, and lanugo hair. Study of fetal cells in amniotic fluid through amniocentesis yields much information about the fetus. Genetic studies (karyotyping) provide knowledge about the sex of the fetus and the number and structure of chromosomes. Other studies such as the lecithin/sphingomyelin (L/S) ratio determine the health or maturity of the fetus.

Yolk Sac

At the same time the amniotic cavity and amnion are forming, another blastocyst cavity forms on the other side of the developing embryonic disk (see Fig. 7-8, B). This cavity becomes surrounded by a membrane, forming the yolk sac. The yolk sac aids in supplying nutrients to the embryo and decidua capsularis by way of the connecting stalk. The inner cell mass of the blastocyst becomes surrounded by a membrane, forming the yolk sac. The yolk sac aids in supplying nutrients to the embryo. Blood and lymphatic vessels develop from the yolk sac as well as the heart, lungs, and liver. The yolk sac is an important structure in early fetal development, playing a role in nutrient supply and blood formation.

Amniotic fluid continues to increase in volume as the fetus grows. At approximately 12 weeks of gestation, the amniotic fluid volume reaches its peak, with a typical range of 800 to 1200 ml. This volume is maintained throughout the remainder of pregnancy, with slight fluctuations occurring due to factors such as fetal movement and swallowing.

The amniotic fluid serves many functions for the embryo/fetus. Amniotic fluid helps maintain a constant body temperature. It serves as a source of oral fluid and as a repository for waste. It cushions the fetus from trauma by blunting and dispersing outside forces. It allows freedom of movement for musculoskeletal development. The fluid keeps the embryo from tangling with the membranes, facilitating symmetric growth of the fetus. If the embryo does become tangled with the membranes, amputations of extremities or other deformities can occur from constricting amniotic bands.

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comes compressed from both sides by the amnion and forms the narrower umbilical cord (see Fig. 7-9). Two arteries carry blood to the chorionic villi from the embryo, and one vein returns blood to the embryo. Approximately 1% of umbilical cords contain only two vessels: one artery and one vein. This occurrence is sometimes associated with congenital malformations.

The cord rapidly increases in length. At term the cord is 2 cm in diameter and ranges from 30 to 90 cm in length (with an average of 55 cm). It twists spirally on itself and loops around the embryo and fetus. A true knot is rare, but false knots occur as folds or kinks in the cord and may jeopardize circulation to the fetus. Connective tissue called Wharton’s jelly prevents compression of the blood vessels and ensures continued nourishment of the embryo and fetus. Compression can occur if the cord lies between the fetal head and the pelvis or is twisted around the fetal body. When the cord is wrapped around the fetal neck, it is called a nuchal cord.

Because the placenta develops from the chorionic villi, the umbilical cord is usually located centrally. A peripheral location is less common and is known as a battledore placenta. The blood vessels are arrayed out from the center to all parts of the placenta.

**Placenta Structure**

The placenta begins to form at implantation. During the third week after conception the trophoblast cells of the chorionic villi continue to invade the decidua basalis. As the uterine capillaries are tapped, the endometrial spiral arteries fill with maternal blood. The chorionic villi grow into the spaces with two layers of cells: the outer syncytium and the inner cytotrophoblast. A third layer develops into anchoring septa, dividing the projecting decidua into separate areas called cotyledons. In each of the 15 to 20 cotyledons, the chorionic villi branch out, and a complex system of fetal blood vessels forms. Each cotyledon is a functional unit. The whole structure is the placenta (Fig. 7-10).

The maternal-placental-embryonic circulation is in place by day 17, when the embryonic heart starts beating. By the end of the third week, embryonic blood is circulating between the embryo and the chorionic villi. In the intervillous spaces, maternal blood supplies oxygen and nutrients to the embryonic capillaries in the villi (Fig. 7-11). Waste products and carbon dioxide diffuse into the maternal blood.

The placenta functions as a means of metabolic exchange. Exchange is minimal at this time because the two cell layers of the villous membrane are too thick. Permeability increases as the cytotrophoblast thins and disappears; by the fifth month, only the single layer of syncytium is left between the maternal blood and the fetal capillaries. The syncytium is the functional layer of the placenta. By the eighth week, genetic testing may be done on a sample of chorionic villi obtained by aspiration biopsy; however, limb defects have been associated with chorionic villus sampling done...
hormone progesterone than the corpus luteum does during development to prepare for lactation. These actions increase the resistance to insulin, facilitate glucose transport, and create placental lactogen (hPL). This substance is similar to human chorionic somatomammotropin (hCS) or human placental lactogen (hPL). It can be detected in the maternal serum by 7 to 10 days after conception, shortly after implantation. This hormone maintains the pregnancy and supports development of the maternal and fetal blood and fetal blood. In this way the placenta functions as a lung for the fetus.

Functions

One of the early functions of the placenta is as an endocrine gland that produces four hormones necessary to maintain the pregnancy and support the embryo/fetus. The hormones are produced in the syncytiotrophoblast. The protein hormone human chorionic gonadotropin (hCG) can be detected in the maternal serum by 7 to 10 days after conception, shortly after implantation. This hormone is the basis for pregnancy tests. The hCG preserves the function of the ovarian corpus luteum, ensuring a continued supply of estrogen and progesterone needed to maintain the pregnancy. Miscarriage occurs if the corpus luteum stops functioning before the placenta can produce sufficient estrogen and progesterone. The hCG reaches its maximum level at 50 to 70 days, then begins to decrease.

The other protein hormone produced by the placenta is human chorionic somatomammotropin (hCS) or human placental lactogen (hPL). This substance is similar to a growth hormone and stimulates maternal metabolism to supply needed nutrients for fetal growth. This hormone increases resistance to insulin, facilitates glucose transport across the placental membrane, and stimulates breast development to prepare for lactation.

The placenta eventually produces more of the steroid hormone progesterone than the corpus luteum does during the first few months of pregnancy. Progesterone maintains the endometrium, decreases the contractility of the uterus, and stimulates development of breast alveoli and maternal metabolism.

By 7 weeks after fertilization, the placenta is producing most of the maternal estrogens, which are steroid hormones. The major estrogen secreted by the placenta is estriol, whereas the ovaries produce mostly estradiol. Measuring estriol levels is a clinical assay for placental functioning. Estrogen stimulates uterine growth and uteroplacental blood flow. It causes a proliferation of the vascular tissue and stimulates myometrial contractility. Placental estrogen production increases greatly toward the end of pregnancy. One theory for the cause of the onset of labor is the decrease in circulating levels of progesterone and the increased levels of estrogen.

The metabolic functions of the placenta include respiration, nutrition, excretion, and storage. Oxygen diffuses from the maternal blood across the placental membrane into the fetal blood, and carbon dioxide diffuses in the opposite direction. In this way the placenta functions as a lung for the fetus.

Carbohydrates, proteins, calcium, and iron are stored in the placenta for ready access to meet fetal needs. Water, inorganic salts, carbohydrates, proteins, fats, and vitamins pass from the maternal blood supply across the placental membrane into the fetal blood, supplying nutrition. Water and electrolytes with a molecular weight less than 500 readily diffuse through the membrane. Hydrostatic and osmotic pressures aid in the flow of water and some solutions. Facilitated and active transport assists in the transfer of glucose, amino acids, calcium, iron, and substances with higher molecular weights. Amino acids and calcium are transported against the concentration gradient between the maternal blood and fetal blood.

The fetal concentration of glucose is lower than the glucose level in the maternal blood because of its rapid metabolism by the fetus. This rapid requirement demands larger concentrations of glucose than simple diffusion can provide. Therefore maternal glucose moves into the fetal circulation by active transport.

Pinocytosis is a mechanism used for transferring large molecules such as albumin and gamma globulins across the placental membrane. This mechanism conveys the maternal immunoglobulins that provide early passive immunity to the fetus.

Metabolic waste products of the fetus cross the placental membrane from the fetal blood into the maternal blood. The maternal kidneys then excrete them. Many viruses can cross the placental membrane and infect the fetus. Some bacteria and protozoa first infect the placenta and then infect the fetus. Drugs can also cross the placental membrane and may harm the fetus. Caffeine, alcohol, nicotine, carbon monoxide and other toxic substances in cigarette smoke, and prescription and recreational drugs (such as marijuana and cocaine) readily cross the placenta (Box 7-4).
Developmentally Toxic Exposures in Humans

<table>
<thead>
<tr>
<th>Substance/Drug</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aminopterin</td>
<td>Lead</td>
</tr>
<tr>
<td>Androgens</td>
<td>Lithium</td>
</tr>
<tr>
<td>Angiotensin-converting enzyme inhibitors</td>
<td>Methyl mercury</td>
</tr>
<tr>
<td>Carbamazepine</td>
<td>Penicillamine</td>
</tr>
<tr>
<td>Cocaine</td>
<td>Phenyltoin</td>
</tr>
<tr>
<td>Coumarin anticoagulants</td>
<td>Radiodine</td>
</tr>
<tr>
<td>Cytomegalovirus</td>
<td>Rubella</td>
</tr>
<tr>
<td>Diethylstilbestrol</td>
<td>Syphilis</td>
</tr>
<tr>
<td>Ethanol (&gt;1 drink/day)</td>
<td>Tetraacycline</td>
</tr>
<tr>
<td>Etretinate</td>
<td>Thalidomide</td>
</tr>
<tr>
<td>Hyperthermia</td>
<td>Toxoplasmosis</td>
</tr>
<tr>
<td>Iodides</td>
<td>Trimethadione</td>
</tr>
<tr>
<td>Ionizing radiation (&gt;10 rad)</td>
<td>Valproic acid</td>
</tr>
<tr>
<td>Isotretinoin</td>
<td>Varicella</td>
</tr>
</tbody>
</table>

Although no direct link exists between the fetal blood in the vessels of the chorionic villi and the maternal blood in the intervillous spaces, only one cell layer separates them. Breaks occasionally occur in the placental membrane. Fetal erythrocytes then leak into the maternal circulation, and the mother may develop antibodies to the fetal red blood cells. This is often the way the Rh-negative mother becomes sensitized to the erythrocytes of her Rh-positive fetus (see the discussion of isoimmunization in Chapter 27).

Though the placenta and fetus are living tissue transplants, they are not destroyed by the host mother (Silver, Peltier, & Branch, 2004). Either the placental hormones suppress the immunologic response, or the tissue evokes no response.

Placental function depends on the maternal blood pressure supplying the circulation. Maternal arterial blood, under pressure in the small uterine spiral arteries, spurs into the intervillous spaces (see Fig. 7.11). As long as rich arterial blood continues to be supplied, pressure is exerted on the blood already in the intervillous spaces, pushing it toward drainage by the low-pressure uterine veins. At term gestation, 10% of the maternal cardiac output goes to the uterus.

If there is interference with the circulation to the placenta, the placenta cannot supply the embryo or fetus. Vasocostriction, such as that caused by hypertension or cocaine use, diminishes uterine blood flow. Decreased maternal blood pressure or decreased cardiac output also diminishes uterine blood flow.

When a woman lies on her back with the pressure of the uterus compressing the vena cava, blood return to the right atrium is diminished (see the discussion of supine hypotension in Chapter 8 and Fig. 14-5). Excessive maternal exercise that diverts blood to the muscles away from the uterus compromises placental circulation. Optimum circulation is achieved when the woman is lying at rest on her side. Decreased uterine circulation may lead to intrapartum growth restriction of the fetus and infants who are small for gestational age.

Braxton Hicks contractions seem to enhance the movement of blood through the intervillous spaces, aiding placental circulation. However, prolonged contractions or too-short intervals between contractions during labor can reduce the blood flow to the placenta.

Fetal Maturation

The stage of the fetus lasts from 9 weeks (when the embryo becomes recognizable as a human being) until the pregnancy ends. Changes during the fetal period are not as dramatic, because refinement of structure and function is taking place. The fetus is less vulnerable to teratogens, except for those that affect central nervous system functioning.

Viability refers to the capability of the fetus to survive outside the uterus. In the past the earliest age at which fetal survival could be expected was 28 weeks after conception. With modern technology and advances in maternal and neonatal care, viability is now possible about 20 weeks after conception (22 weeks since LMP, fetal weight of 500 g or more).

The limitations on survival outside the uterus are based on central nervous system function and oxygenation capability of the lungs.

Respiratory System

The respiratory system begins development during embryonic life and continues through fetal life and into childhood. The development of the respiratory tract begins in week 4 and continues through week 17 with formation of the trachea, bronchi, and lung buds. Between 16 and 24 weeks the bronchi and terminal bronchioles enlarge, and vascular structures and primitive alveoli are formed. Between 24 weeks and term birth, more alveoli form. Specialized alveolar cells, type I and type II cells, secrete pulmonary surfactants to line the interior of the alveoli. After 12 weeks, sufficient surfactant is present in developed alveoli to provide infants with a good chance of survival.

Pulmonary surfactants. The detection of the presence of pulmonary surfactants (surface-active phospholipids) in amniotic fluid has been used to determine the degree of fetal lung maturity, or the ability of the lungs to function after birth. Lecithin (L) is the most critical alveolar surfactant required for postnatal lung expansion. It is detectable at approximately 21 weeks and increases in amount after week 24. Another pulmonary phospholipid, sphingomyelin (S), remains constant in amount. Therefore the measure of lecithin in relation to sphingomyelin, or the L/S ratio, is used to determine fetal lung maturity. When the L/S ratio reaches 2.1, the infant’s lungs are considered to be mature. This occurs at approximately 36 weeks of gestation (Mercer, 2004).

Certain maternal conditions that cause decreased maternal placental blood flow, such as maternal hypertension, placental dysfunction, infection, or corticosteroid use, accelerate lung maturity. This apparently is caused by the

**BOX 7-4 Developmentally Toxic Exposures in Humans**
resulting fetal hypoxia, which stresses the fetus and increases the blood levels of corticosteroids that accelerate alveolar and surfactant development.

Conditions such as gestational diabetes and chronic glomerulonephritis can retard fetal lung maturity. The use of intrabronchial synthetic surfactant in the treatment of respiratory distress syndrome in the newborn has greatly improved the chances of survival for preterm infants.

Fetal respiratory movements have been seen on ultrasound as early as the eleventh week. These fetal respiratory movements may aid in development of the chest wall muscles and regulate lung fluid volume. The fetal lungs produce fluid that expands the air spaces in the lungs. The fluid drains into the amniotic fluid or is swallowed by the fetus.

Before birth, secretion of lung fluid decreases. The normal birth process squeezes out approximately one third of the fluid. Infants of cesarean births do not benefit from this squeezing process; therefore they may have more respiratory difficulty at birth. The fluid remaining in the lungs at birth is usually reabsorbed into the infant's bloodstream within 2 hours of birth.

**Fetal circulatory system**

The cardiovascular system is the first organ system to function in the developing human. Blood vessel and blood cell formation begins in the third week and supplies the embryo with oxygen and nutrients from the mother. By the end of the third week the tubular heart begins to beat, and the primitive cardiovascular system links the embryo, connecting stalk, chorion, and yolk sac. During the fourth and fifth weeks the heart develops into a four-chambered organ. By the end of the embryonic stage, the heart is developmentally complete.

The fetal lungs do not function for respiratory gas exchange, so a special circulatory pathway, the ductus arteriosus, bypasses the lungs. Oxygen-rich blood from the placenta flows rapidly through the umbilical vein into the fetal abdomen (Fig. 7-12). When the umbilical vein reaches the liver, it divides into two branches. One branch circulates some oxygenated blood through the liver. Most of the blood passes through the ductus venosus into the inferior vena cava. There it mixes with the deoxygenated blood from the fetal legs and abdomen on its way to the right atrium. Most of this blood passes straight through the right atrium and
through the foramen ovale, an opening into the left atrium. There it mixes with the small amount of deoxygenated blood returning from the fetal lungs through the pulmonary veins.

The blood flows into the left ventricle and is squeezed out into the aorta, where the arteries supplying the heart, head, neck, and arms receive most of the oxygen-rich blood. This pattern of supplying the highest levels of oxygen and nutrients to the head, neck, and arms enhances the cephalocaudal (head-to-rump) development of the embryo and fetus.

Deoxygenated blood returning from the head and arms enters the right atrium through the superior vena cava. This blood is directed downward into the right ventricle, where it is squeezed into the pulmonary artery. A small amount of blood circulates through the resistant lung tissue, but the majority follows the path with less resistance through the ductus arteriosus into the aorta, distal to the point of exit of the arteries supplying the head and arms with oxygenated blood. The oxygen-poor blood flows through the abdominal aorta into the internal iliac arteries, where the umbilical arteries direct most of it back through the umbilical cord to the placenta. There the blood gives up its wastes and carbon dioxide in exchange for nutrients and oxygen. The blood remaining in the iliac arteries flows through the fetal abdomen and legs, ultimately returning through the inferior vena cava to the heart.

The following three special characteristics enable the fetus to obtain sufficient oxygen from the maternal blood:
- Fetal hemoglobin carries 20% to 30% more oxygen than maternal hemoglobin.
- The hemoglobin concentration of the fetus is about 50% greater than that of the mother.
- The fetal heart rate (FHR) is 110 to 160 beats/min, making the cardiac output per unit of body weight higher than that of an adult.

**Hematopoietic system**

Hematopoiesis, the formation of blood, occurs in the yolk sac (see Fig. 7-8, B) beginning in the third week. Hematopoietic stem cells seed the fetal liver during the fifth week, and hematopoiesis begins there during the sixth week. The antigenic factors that determine blood type are present in the erythrocytes soon after the sixth week. For this reason the Rh-negative woman is at risk for isoimmunization in any pregnancy that lasts longer than 6 weeks after fertilization.

**Hepatic system**

The liver and biliary tract develop from the foregut during the fourth week of gestation. Hematopoiesis begins during the sixth week and requires that the liver be large. The embryonic liver is prominent, occupying most of the abdominal cavity. Bile, a constituent of meconium, begins to form in the twelfth week.

Glycogen is stored in the fetal liver beginning at week 9 or 10. At term, glycogen stores are twice those of the adult. Glycogen is the major source of energy for the fetus and for the neonate stressed by in utero hypoxia, extraterine loss of the maternal glucose supply, the work of breathing, or cold stress. Iron is also stored in the fetal liver. If maternal intake is sufficient, the fetus can store enough iron to last for 5 months after birth.

During fetal life the liver does not have to conjugate bilirubin for excretion because the unconjugated bilirubin is cleared by the placenta. Therefore the glucuronol trans-

ferase enzyme needed for conjugation is present in the fe-
tal liver in amounts less than those required after birth. This predisposes the neonate, especially the preterm infant, to hyperbilirubinemia.

Coagulation factors II, VII, IX, and X cannot be syn-
thesized in the fetal liver because of the lack of vitamin K synthesis in the sterile fetal gut. This coagulation deficiency persists after birth for several days and is the rationale for the prophylactic administration of vitamin K to the newborn.

**Gastrointestinal system**

During the fourth week the shape of the embryo changes from almost straight to a C shape as both ends fold in toward the ventral surface. A portion of the yolk sac is incor-
portated into the body from head to tail as the primitive gut (digestive system).

The foregut produces the pharynx, part of the lower res-
piratory tract, the esophagus, the stomach, the first half of the duodenum, the liver, the pancreas, and the gallbladder. These structures evolve during the fifth and sixth weeks. Malformations that can occur in these areas include esophageal atresia, hypertrophic pyloric stenosis, duodenal stenosis or atresia, and biliary atresia.

The midgut becomes the distal half of the duodenum, the jejunum and ileum, the cecum and appendix, and the proximal half of the colon. The midgut loop projects into the umbilical cord between weeks 5 and 10. A malformation (or omphalocele) results if the midgut fails to return to the ab-
dominal cavity, causing the intestines to protrude from the umbilicus. Meckel’s diverticulum is the most common mal-
formation of the midgut. It occurs when a remnant of the yolk stalk that has failed to degenerate attaches to the ileum, leaving a blind sac.

The hindgut develops into the distal half of the colon, the rectum and parts of the anal canal, the urinary bladder, and the urethra. Anorectal malformations are the most common abnormalities of the digestive system.

The fetus swallows amniotic fluid beginning in the fifth month. Gastric emptying and intestinal peristalsis occur. Pe-
tal nutrition and elimination needs are taken care of by the placenta. As the fetus nears term, fetal waste products ac-
cumulate in the intestines as dark green to black, tarry meco-
nium. Normally this substance is passed through the rectum
within 24 hours of birth. Sometimes with a breech presentation or fetal hyponxia, meconium is passed in utero into the amniotic fluid. The failure to pass meconium after birth may indicate atresia somewhere in the digestive tract; an imperforate anus; or meconium ileus, in which a firm meconium plug blocks passage (seen in infants with CF).

The metabolic rate of the fetus is relatively low, but the infant has great growth and development needs. Beginning in week 9 the fetus synthesizes glycogen for storage in the liver. Between 26 and 30 weeks the fetus begins to lay down stores of brown fat in preparation for extrauterine cold stress. Thermo-regulation in the neonate requires increased metabolism and adequate oxygenation.

The gastrointestinal system is mature by 36 weeks. Digestive enzymes (except pancreatic amylase and lipase) are present in sufficient quantity to facilitate digestion. The neonate cannot digest starches or fats efficiently. Little saliva is produced.

Renal system
The kidneys form during the fifth week and begin to function approximately 4 weeks later. Urine is excreted into the amniotic fluid and forms a major part of the amniotic fluid volume. Oligohydramnios is indicative of renal dysfunction. Because the placenta acts as the organ of excretion and maintains fetal water and electrolyte balance, the fetus does not need functioning kidneys while in utero. At birth, however, the kidneys are required immediately for excretion and acid-base regulatory functions. A fetal renal malformation can be diagnosed in utero. Corrective or palliative fetal surgery may treat the malformation successfully, or plans can be made for treatment immediately after birth.

At term the fetus has fully developed kidneys. However, the glomerular filtration rate (GFR) is low, and the kidneys lack the ability to concentrate urine. This makes the newborn more susceptible to both overhydration and dehydration.

Most newborns void within 24 hours of birth. With the loss of the swallowed amniotic fluid and the metabolism of nutrients provided by the placenta, voidings for the first days of life are scant until fluid intake increases.

Neurologic system
The nervous system originates from the ectoderm during the third week after fertilization. The open neural tube forms during the fourth week. It initially closes at what will be the junction of the brain and spinal cord, leaving both ends open. The embryo folds in on itself lengthwise at this time, forming a head fold in the neural tube at this junction. The cranial end of the neural tube closes, then the caudal end closes. During week 5, different growth rates cause more flexures in the neural tube, delineating three brain areas: the forebrain, midbrain, and hindbrain.

The forebrain develops into the eyes (cranial nerve II) and cerebral hemispheres. The development of all areas of the cerebral cortex continues throughout fetal life and into childhood. The olfactory system (cranial nerve I) and thalamus also develop from the forebrain. Cranial nerves III and IV (oculomotor and trochlear) form from the midbrain. The hindbrain forms the medulla, the pons, the cerebellum, and the remainder of the cranial nerves. Brain waves can be recorded on an electroencephalogram by week 8.

The spinal cord develops from the long end of the neural tube. Another ectodermal structure, the neural crest, develops into the peripheral nervous system. By the eighth week, nerve fibers traverse throughout the body. By week 11 or 12 the fetus makes respiratory movements, moves all extremities, and changes position in utero. The fetus can suck his or her thumb, swim in the amniotic fluid pool, and turn somersaults, and sometimes ties a knot in the umbilical cord.

Box 7-5 describes the major types of fetal movements. Some-time between 16 and 20 weeks, when the movements are strong enough to be perceived by the mother as “the baby moving,” quickening has occurred. The perception of move-ment occurs earlier in the multipara than in the primipara.

**BOX 7-5**

**Major Types of Fetal Movements**

<table>
<thead>
<tr>
<th>General movements</th>
<th>These slow, gross movements involve the whole body. Their duration is from several seconds to a minute.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Startle movements</td>
<td>These quick (less than 1 second), generalized movements always start in the limbs and may spread to the trunk and neck.</td>
</tr>
<tr>
<td>Hiccups</td>
<td>These are repetitive phasic contractions of the diaphragm. A bout may last several minutes.</td>
</tr>
<tr>
<td>Fetal breathing movements</td>
<td>These are paradoxic movements in which the thorax moves inward and the abdomen outward with each contraction of the diaphragm.</td>
</tr>
<tr>
<td>Isolated arm or leg movements</td>
<td>These movements of the extremities occur without movement of the trunk.</td>
</tr>
<tr>
<td>Hand-face contact</td>
<td>This occurs any time the moving hand makes contact with the face or mouth.</td>
</tr>
<tr>
<td>Retroflexion of the head</td>
<td>This is a slow to jerky backward bending of the head.</td>
</tr>
<tr>
<td>Lateral rotation of the head</td>
<td>This involves isolated turning of the head from side to side.</td>
</tr>
<tr>
<td>Antetellexion of the head</td>
<td>This is a normally slow forward bending of the head.</td>
</tr>
<tr>
<td>Opening of the mouth</td>
<td>This isolated movement may be accompanied by protrusion of the tongue.</td>
</tr>
<tr>
<td>Yawn</td>
<td>The mouth is slowly opened and rapidly closed after a few seconds.</td>
</tr>
<tr>
<td>Sucking</td>
<td>This burst of rhythmic jaw movements is sometimes followed by swallowing. With this movement the fetus may be drinking amniotic fluid.</td>
</tr>
<tr>
<td>Stretch</td>
<td>This complex movement involves overextension of the spine, retroflexion of the head, and elevation of the arms.</td>
</tr>
</tbody>
</table>

The mother also becomes aware of the sleep and wake cycles of the fetus.

**Sensory awareness.** Purposeful movements of the fetus have been demonstrated in response to a firm touch transmitted through the mother’s abdomen. Because it can feel, the fetus requires anesthesia when invasive procedures are done. Fetuses respond to sound by 24 weeks. Different types of music evoke different movements. The fetus can be soothed by the sound of the mother’s voice. Acoustic stimulation can be used to evoke an FHR response. The fetus becomes accustomed (habituates) to noises heard repeatedly. Hearing is fully developed at birth.

The fetus is able to distinguish taste. By the fifth month, when the fetus is swallowing amniotic fluid, a sweetener added to the fluid causes the fetus to swallow faster. The fetus also reacts to temperature changes. A cold solution placed into the amniotic fluid can cause fetal hiccups.

The fetus can see. Eyes have both rods and cones in the retina by the seventh month. A bright light shone on the mother’s abdomen in late pregnancy causes abrupt fetal movements. During sleep time, rapid eye movements (REMs) have been observed similar to those occurring in children and adults while dreaming.

At term the fetal brain is approximately one fourth the size of an adult brain. Neurologic development continues. Stressors on the fetus and neonate (e.g., chronic poor nutrition or hypoxia, drugs, environmental toxins, trauma, or disease) cause damage to the central nervous system long after the vulnerable embryonic time for malformations in other organ systems. Neurologic insult can result in cerebral palsy, neuromuscular impairment, mental retardation, and learning disabilities.

**Endocrine system**

The thyroid gland develops along with structures in the head and neck during the third and fourth weeks. The secretion of thyroxine begins during the eighth week. Maternal thyroxine does not readily cross the placenta; therefore the fetus that does not produce thyroid hormones will be born with congenital hypothyroidism. If untreated, hypothyroidism can result in severe mental retardation. Screening for hypothyroidism is typically included in the testing when screening for PKU after birth.

The adrenal cortex is formed during the sixth week and produces hormones by the eighth or ninth week. As term approaches, the fetus produces more cortisol. This is believed to aid in initiation of labor by decreasing the maternal progesterone and stimulating production of prostaglandins.

The pancreas forms from the foregut during the fifth through eighth weeks. The islets of Langerhans develop during the twelfth week. Insulin is produced by the twentieth week. In infants of mothers with uncontrolled diabetes, maternal hyperglycemia produces fetal hyperglycemia, stimulating hyperinsulinemia and islet cell hyperplasia. This results in a macrosomic (large-sized) fetus. The hyperinsulinemia also blocks lung maturation, placing the neonate at risk for respiratory distress and hypoglycemia when the maternal glucose source is lost at birth. Control of the maternal glucose level before and during pregnancy minimizes problems for the fetus and infant.

**Reproductive system**

Sex differentiation begins in the embryo during the seventh week. Distinguishing characteristics appear around the ninth week and are fully differentiated by the twelfth week. When a Y chromosome is present, testes are formed. By the end of the embryonic period, testosterone is being secreted and causes formation of the male genitalia. By week 28 the testes begin descending into the scrotum. After birth, low levels of testosterone continue to be secreted until the pubertal surge.

The female, with two X chromosomes, forms ovaries and female external genitalia. By the sixteenth week, oogenesis has been established. At birth the ovaries contain the female’s lifetime supply of ova. Most female hormone production is delayed until puberty. However, the fetal endometrium responds to maternal hormones, and withdrawal bleeding or vaginal discharge (pseudomenstruation) may occur at birth when these hormones are lost. The high level of maternal estrogen also stimulates mammary engorgement and secretion of fluid (“witch’s milk”) in newborn infants of both sexes.

**Musculoskeletal system**

Bones and muscles develop from the mesoderm by the fourth week of embryonic development. At that time the cardiac muscle is already beating. The mesoderm next to the neural tube forms the vertebral column and ribs. The parts of the vertebral column grow toward each other to enclose the developing spinal cord. Ossification, or bone formation, begins. If there is a defect in the bony fusion, various forms of spina bifida may occur. A large defect affecting several vertebrae may allow the membranes and spinal cord to pouch out from the back, producing neurologic deficits and skeletal deformity. The flat bones of the skull develop during the embryonic period, and ossification continues throughout childhood. At birth, connective tissue sutures exist where the bones of the skull meet. The areas where more than two bones meet (called fontanels) are especially prominent. The sutures and fontanels allow the bones of the skull to mold, or move during birth, enabling the head to pass through the birth canal.

The bones of the shoulders, arms, hips, and legs appear in the sixth week as a continuous skeleton with no joints. Differentiation occurs, producing separate bones and joints. Ossification will continue through childhood to allow growth. Beginning in the seventh week, muscles contract spontaneously. Arm and leg movements are visible on ultrasound, although the mother does not perceive them until sometime between 16 and 20 weeks.
Integumentary system

The epidermis begins as a single layer of cells derived from the ectoderm at 4 weeks. By the seventh week, there are two layers of cells. The cells of the superficial layer are sloughed and become mixed with the sebaceous gland secretions to form the white, cheesy vernix caseosa, the material that protects the skin of the fetus. The vernix is thick at 24 weeks but becomes scant by term.

The basal layer of the epidermis is the germinal layer, which replaces lost cells. Until 17 weeks the skin is thin and wrinkled, with blood vessels visible underneath. The skin thickens, and all layers are present at term. After 32 weeks, as subcutaneous fat is deposited under the dermis, the skin becomes less wrinkled and red in appearance.

By 16 weeks the epidermal ridges are present on the palms of the hands, the fingers, the bottom of the feet, and the toes. These handprints and footprints are unique to that infant.

Hairs form from hair bulbs in the epidermis that project into the dermis. Cells in the hair bulb keratinize to form the hair shaft. As the cells at the base of the hair shaft proliferate, the hair grows to the surface of the epithelium. Very fine hairs, called lanugo, appear first at 12 weeks on the eyebrows and upper lip. By 20 weeks they cover the entire body. At this time the eyelashes, eyebrows, and scalp hair are beginning to grow. By 28 weeks the scalp hair is longer than the lanugo, which thins and may disappear by term gestation.

Fingernails and toenails develop from thickened epidermis at the tips of the digits beginning during the tenth week. They grow slowly. Fingernails usually reach the fingertips by 32 weeks, and toenails reach the toes by 36 weeks.

Immunologic system

During the third trimester, albumin and globulin are present in the fetus. The only immunoglobulin (Ig) that crosses the placenta, IgG, provides passive acquired immunity to specific bacterial toxins. The fetus produces IgM immunoglobulins by the end of the first trimester. These are produced in response to blood group antigens, gram-negative enteric organisms, and some viruses. IgA immunoglobulins are not produced by the fetus; however, colostrum, the precursor to breast milk, contains large amounts of IgA and can provide passive immunity to the neonate who is breastfed (Table 7-1).

The normal term neonate can fight infection, but not as effectively as an older child. The preterm infant is at much greater risk for infection.

Table 7-1 summarizes embryonic and fetal development.

Multifetal Pregnancy

Twins

The incidence of twinning is 1 in 43 pregnancies (Benirschke, 2004). There has been a steady rise in multiple births since 1973. This is partly attributed to delayed childbearing. The use of ovulation-enhancing drugs is also a factor.

Dizygotic twins. When two mature ova are produced in one ovarian cycle, both have the potential to be fertilized by separate sperm. This results in two zygotes, or dizygotic twins (Fig. 7-13). There are always two amnions, two chorions, and two placentas that may be fused together. These dizygotic fraternal twins may be the same sex or different sexes and are genetically no more alike than siblings born at different times. Dizygotic twinning occurs in families, is more common among African-American women than Caucasian women, and is least common among Asian-American women. Dizygotic twinning increases in frequency with maternal age up to 35 years, with parity, and with the use of fertility drugs.

Monozygotic twins. Identical or monozygotic twins develop from one fertilized ovum, which then divides (Fig. 7-14). They are the same sex and have the same genotype. If division occurs soon after fertilization, two embryos, two amnions, two chorions, and two placentas that may be fused will develop. Most often, division occurs between 4 and 8 days after fertilization, and there are two embryos.

<table>
<thead>
<tr>
<th>CLASS</th>
<th>LOCATION</th>
<th>CHARACTERISTICS</th>
</tr>
</thead>
<tbody>
<tr>
<td>IgG</td>
<td>Plasma, interstitial fluid</td>
<td>Is only immunoglobulin that crosses placenta</td>
</tr>
<tr>
<td>IgA</td>
<td>Body secretions, including tears, saliva, breast milk, colostrum</td>
<td>Is responsible for secondary immune response</td>
</tr>
<tr>
<td>IgM</td>
<td>Plasma</td>
<td>Lines mucous membranes and protects body surfaces</td>
</tr>
<tr>
<td>IgD</td>
<td>Plasma</td>
<td>Is responsible for primary immune response</td>
</tr>
<tr>
<td>IgE</td>
<td>Plasma, interstitial fluids</td>
<td>Causes symptoms of allergic reactions</td>
</tr>
</tbody>
</table>

Fig. 7-13  Formation of dizygotic twins. There is fertilization of two ova, two implantations, two placentas, two chorions, and two amnions.

Two amnions, two chorions, and one placenta. Rarely, division occurs after the eighth day following fertilization. In this case there are two embryos within a common amnion and a common chorion with one placenta. This often causes circulatory problems because the umbilical cords may tangle together, and one or both fetuses may die. If division occurs very late, cleavage may not be complete, and conjoined or “Siamese” twins could result. Monozygotic twinning occurs in approximately 1 of 250 births (Benirschke, 2004). There is no association with race, heredity, maternal age, or parity. Fertility drugs increase the incidence of monozygotic twinning.

Other multifetal pregnancies

The occurrence of multifetal pregnancies with three or more fetuses has increased with the use of fertility drugs and IVF. Triplets occur in about 1 of 1341 pregnancies (Benirschke, 2004). They can occur from the division of one zygote into two, with one of the two dividing again, producing identical triplets. Triplets can also be produced from two zygotes, one dividing into a set of identical twins and the second zygote a single fraternal sibling, or from three zygotes. Quadruplets, quintuplets, sextuplets, and so on have similar possible derivations.

Fig. 7-14  Formation of monozygotic twins.  
A. One fertilization: blastomeres separate, resulting in two implantations, two placentas, and two sets of membranes.  
B. One blastomere with two inner cell masses, one fused placenta, one chorion, and separate amnions.  
C. One blastomere with incomplete separation of cell mass resulting in conjoined twins.
# TABLE 7-2

Milestones in Human Development before Birth since Last Menstrual Period

<table>
<thead>
<tr>
<th>4 WEEKS</th>
<th>8 WEEKS</th>
<th>12 WEEKS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>EXTERNAL APPEARANCE</strong></td>
<td>Body fairly well formed; nose flat, eyes far apart; digits well formed; head elevating; tail almost disappeared; eyes, ears, nose, and mouth recognizable.</td>
<td>Nails appearing; resembles a human; head erect but disproportionately large; skin pink, delicate.</td>
</tr>
<tr>
<td><strong>CROWN-TO-RUMP MEASUREMENT; WEIGHT</strong></td>
<td>2.5-3 cm; 2 g</td>
<td>6-9 cm; 19 g</td>
</tr>
<tr>
<td><strong>GASTROINTESTINAL SYSTEM</strong></td>
<td>Intestinal villi developing; small intestines coil within umbilical cord; palatal folds present; liver very large.</td>
<td>Bile secreted; palatal fusion complete; intestines have withdrawn from cord and assume characteristic positions.</td>
</tr>
<tr>
<td><strong>ALL SOMITES PRESENT</strong></td>
<td>First indication of ossification—occiput, mandible, and humerus; fetus capable of some movement; definitive muscles of trunk, limbs, and head well represented.</td>
<td>Some bones well outlined, ossification spreading; upper cervical to lower sacral arches and bodies ossify; smooth muscle layers indicated in hollow viscera.</td>
</tr>
<tr>
<td><strong>CIRCULATORY SYSTEM</strong></td>
<td>Main blood vessels assume final plan; enucleated red cells predominate in blood.</td>
<td>Blood forming in marrow.</td>
</tr>
<tr>
<td><strong>RESPIRATORY SYSTEM</strong></td>
<td>Pleural and pericardial cavities forming; branching bronchioles; nostrils closed by epithelial plugs.</td>
<td>Lungs acquire definite shape; vocal cords appear.</td>
</tr>
<tr>
<td><strong>RENA L SYSTEM</strong></td>
<td>Earliest secretory tubules differentiating; bladder-urethra separates from rectum.</td>
<td>Kidney able to secrete urine; bladder expands as a sac.</td>
</tr>
<tr>
<td><strong>NERVOUS SYSTEM</strong></td>
<td>Cerebral cortex begins to acquire typical cells; differentiation of cerebral cortex, meninges, ventricular foramina, cerebrospinal fluid circulation; spinal cord extends entire length of spine.</td>
<td>Brain structural configuration almost complete; cord shows cervical and lumbar enlargements; fourth ventricle foramina are developed; sucking present.</td>
</tr>
<tr>
<td><strong>SENSORY ORGANS</strong></td>
<td>Primordial choroid plexuses develop; ventricles large relative to cortex; development progressing; eyes converging rapidly; internal ear developing; eyelids fuse.</td>
<td>Earliest taste buds indicated; characteristic organization of eye attained.</td>
</tr>
<tr>
<td><strong>GENITAL SYSTEM</strong></td>
<td>Testes and ovaries distinguishable; external genitalia sexless but begin to differentiate.</td>
<td>Sex recognizable; internal and external sex organs specific.</td>
</tr>
</tbody>
</table>

**Table 7-2**

**Milestones in Human Development before Birth since Last Menstrual Period**

<table>
<thead>
<tr>
<th>Milestone</th>
<th>4 Weeks</th>
<th>8 Weeks</th>
<th>12 Weeks</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Body</strong></td>
<td>Flexed, C shaped; arm and leg buds present; head at right angles to body.</td>
<td>Body fairly well formed; nose flat, eyes far apart; digits well formed; head elevating; tail almost disappeared; eyes, ears, nose, and mouth recognizable.</td>
<td>Nails appearing; resembles a human; head erect but disproportionately large; skin pink, delicate.</td>
</tr>
<tr>
<td><strong>Crown-to-Rump Measurement</strong></td>
<td>0.4-0.5 cm; 0.4 g</td>
<td>2.5-3 cm; 2 g</td>
<td>6-9 cm; 19 g</td>
</tr>
<tr>
<td><strong>Gastrointestinal System</strong></td>
<td>Stomach at midline and fusiform; conspicuous liver; esophagus short; intestines a short tube.</td>
<td>Intestinal villi developing; small intestines coil within umbilical cord; palatal folds present; liver very large.</td>
<td>Bile secreted; palatal fusion complete; intestines have withdrawn from cord and assume characteristic positions.</td>
</tr>
<tr>
<td><strong>Musculoskeletal System</strong></td>
<td>All somites present.</td>
<td>First indication of ossification—occiput, mandible, and humerus; fetus capable of some movement; definitive muscles of trunk, limbs, and head well represented.</td>
<td>Some bones well outlined, ossification spreading; upper cervical to lower sacral arches and bodies ossify; smooth muscle layers indicated in hollow viscera.</td>
</tr>
<tr>
<td><strong>Circulatory System</strong></td>
<td>Heart develops, double chambers visible; begins to beat; aortic arch and major veins formed.</td>
<td>Main blood vessels assume final plan; enucleated red cells predominate in blood.</td>
<td>Blood forming in marrow.</td>
</tr>
<tr>
<td><strong>Respiratory System</strong></td>
<td>Primary lung buds appear.</td>
<td>Pleural and pericardial cavities forming; branching bronchioles; nostrils closed by epithelial plugs.</td>
<td>Lungs acquire definite shape; vocal cords appear.</td>
</tr>
<tr>
<td><strong>Renal System</strong></td>
<td>Rudimentary urethral buds appear.</td>
<td>Earliest secretory tubules differentiating; bladder-urethra separates from rectum.</td>
<td>Kidney able to secrete urine; bladder expands as a sac.</td>
</tr>
<tr>
<td><strong>Nervous System</strong></td>
<td>Well-marked midbrain flexure; no hindbrain or cervical flexures; neural groove closed.</td>
<td>Cerebral cortex begins to acquire typical cells; differentiation of cerebral cortex, meninges, ventricular foramina, cerebrospinal fluid circulation; spinal cord extends entire length of spine.</td>
<td>Brain structural configuration almost complete; cord shows cervical and lumbar enlargements; fourth ventricle foramina are developed; sucking present.</td>
</tr>
<tr>
<td><strong>Sensory Organs</strong></td>
<td>Eye and ear appearing as optic vesicle and otocyst.</td>
<td>Primordial choroid plexuses develop; ventricles large relative to cortex; development progressing; eyes converging rapidly; internal ear developing; eyelids fuse.</td>
<td>Earliest taste buds indicated; characteristic organization of eye attained.</td>
</tr>
<tr>
<td><strong>Genital System</strong></td>
<td>Genital ridge appears (fifth week).</td>
<td>Testes and ovaries distinguishable; external genitalia sexless but begin to differentiate.</td>
<td>Sex recognizable; internal and external sex organs specific.</td>
</tr>
</tbody>
</table>
### TABLE 7-2

**Milestones in Human Development before Birth since Last Menstrual Period—cont’d**

<table>
<thead>
<tr>
<th>16 WEEKS</th>
<th>20 WEEKS</th>
<th>24 WEEKS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>EXTERNAL APPEARANCE</strong></td>
<td>Head still dominant; face looks human; eyes, ears, and nose approach typical appearance on gross examination; arm/leg ratio proportionate; scalp hair appears</td>
<td>Vernix caseosa appears; lanugo appears; legs lengthen considerably; sebaceous glands appear</td>
</tr>
<tr>
<td><strong>CROWN-TO-RUMP MEASUREMENT; WEIGHT</strong></td>
<td>11.5-13.5 cm; 100 g</td>
<td>16-18.5 cm; 300 g</td>
</tr>
<tr>
<td><strong>GASTROINTESTINAL SYSTEM</strong></td>
<td>Meconium in bowel; some enzyme secretion; anus open</td>
<td>Enamel and dentine depositing; ascending colon recognizable</td>
</tr>
<tr>
<td><strong>MUSCULOSKELETAL SYSTEM</strong></td>
<td>Most bones distinctly indicated throughout body; joint cavities appear; muscular movements can be detected</td>
<td>Sternum ossifies; fetal movements strong enough for mother to feel</td>
</tr>
<tr>
<td><strong>CIRCULATORY SYSTEM</strong></td>
<td>Heart muscle well developed; blood formation active in spleen</td>
<td>Blood formation increases in bone marrow and decreases in liver</td>
</tr>
<tr>
<td><strong>RESPIRATORY SYSTEM</strong></td>
<td>Elastic fibers appears in lungs; terminal and respiratory bronchioles appear</td>
<td>Nostrils reopen; primitive respiratory-like movements begin</td>
</tr>
<tr>
<td><strong>RENAL SYSTEM</strong></td>
<td>Kidney in position; attains typical shape and plan</td>
<td></td>
</tr>
<tr>
<td><strong>NERVOUS SYSTEM</strong></td>
<td>Cerebral lobes delineated; cerebellum assumes some prominence</td>
<td>Brain grossly formed; cord myelination begins; spinal cord ends at level of first sacral vertebra (S1)</td>
</tr>
<tr>
<td><strong>SENSORY ORGANS</strong></td>
<td>General sense organs differentiated</td>
<td>Nose and ears ossify</td>
</tr>
<tr>
<td><strong>GENITAL SYSTEM</strong></td>
<td>Testes in position for descent into scrotum; vagina open</td>
<td></td>
</tr>
</tbody>
</table>
## TABLE 7-2

<table>
<thead>
<tr>
<th>Milestones in Human Development before Birth since Last Menstrual Period—cont’d</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>28 WEEKS</strong></td>
</tr>
<tr>
<td><strong>EXTERNAL APPEARANCE</strong></td>
</tr>
<tr>
<td>Lean body, less wrinkled and red; nails appear</td>
</tr>
<tr>
<td><strong>CROWN-TO-RUMP MEASUREMENT; WEIGHT</strong></td>
</tr>
<tr>
<td><strong>MUSCULOSKELETAL SYSTEM</strong></td>
</tr>
<tr>
<td>Astragalus (talus, ankle bone) ossifies; weak, fleeting movements, minimum tone</td>
</tr>
<tr>
<td><strong>RESPIRATORY SYSTEM</strong></td>
</tr>
<tr>
<td>Lecithin forming on alveolar surfaces</td>
</tr>
<tr>
<td><strong>RENAL SYSTEM</strong></td>
</tr>
<tr>
<td><strong>NERVOUS SYSTEM</strong></td>
</tr>
<tr>
<td>Appearance of cerebral fissures, convolutions rapidly appearing; indeterminate sleep-wake cycle; cry weak or absent; weak suck reflex</td>
</tr>
<tr>
<td><strong>SENSORY ORGANS</strong></td>
</tr>
<tr>
<td>Eyelids reopen; retinal layers completed, light receptive; pupils capable of reacting to light</td>
</tr>
<tr>
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</tr>
<tr>
<td></td>
</tr>
</tbody>
</table>
COMMUNITY ACTIVITY

Select two Web addresses for resources on genetics for parents from the Resource list provided in this chapter. Access the sites.

Compare and contrast the appearance, the readability, and the information contained in the sites.

To whom would you recommend these sites?
Is the information culturally relevant?
What information would parents need?
How could you as a nurse use this information?

Key Points

- Genetic disease affects people of all ages, from all socioeconomic levels, and from all racial and ethnic backgrounds.
- Genetic disorders span every clinical practice specialty.
- Nurses with advanced preparation are assuming important roles in genetic counseling.
- Genes are the basic units of heredity, responsible for all human characteristics. They make up 23 pairs of chromosomes: 22 pairs of autosomes and one pair of sex chromosomes.
- Genetic disorders follow mendelian inheritance patterns of dominance, segregation, and independent assortment of normal genetic transmission.
- Multifactorial inheritance includes both genetic and environmental contributions.
- Human gestation is approximately 280 days after the LMP, or 266 days after conception.
- Multifactorial inheritance includes both genetic and environmental contributions.

Answer Guidelines to Critical Thinking Exercise

Ultrasound Dating of Pregnancy

1. Yes, ultrasound dating of pregnancies is well established. The nurse can use photographs or drawings of a fetus to educate Adrienne about the appearance of a fetus at different gestational ages. In addition, charts of embryonic development including critical periods of development can be used.

2. The nurse can assume the following:
   a. Adrienne wants to learn about fetal development.
   b. Depending on prior education, Adrienne may know little about embryonic and fetal development.
   c. Adrienne has the right to know the risks and benefits of ultrasound testing and to give informed consent for the procedure.
   d. Dating the pregnancy is important to identify and prepare for variations from normal.

3. Priorities for nursing care include educating Adrienne about fetal development, ensuring that she is aware of the risks and benefits of ultrasound testing, obtaining a signature for the procedure on a consent form (if her physician has not already done so), informing Adrienne what ultrasound testing entails, and providing support as necessary during the procedure.

4. Yes, ultrasound testing can provide accurate dating of a pregnancy and allow detection of structural anomalies in a fetus.

5. Adrienne has the right to refuse to learn about fetal development and to refuse ultrasound testing. She may or may not want to learn the sex of the fetus; her wishes should be respected in this regard.

Resources

Alliance of Genetic Support Groups
www.geneticalliance.org

Ask NOAH About: Pregnancy
www.nouch.cuny.edu/pregnancy/pregnancy.html

Family Guide to Cystic Fibrosis Genetic Testing
www.phd.msu.edu/cf/fam.html

GeneClinics
www.geneclinics.org

GeneTests
www.herb.washington.edu/helix

Genetics & Ethics
www.genetics.ca/

Genetics Education for Nurses
www.cincinnatichildrens.org/ed/clinical/gennet/default.htm

Genetics Home Reference
www.ghr.nlm.nih.gov/

Information for Genetic Professionals
www.kumc.edu/gpo/geneinfo.html

International Society of Nurses in Genetics (ISONG)
www.nursing.creighton.edu/isong

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GeneClinics
www.geneclinics.org

GeneTests
www.herb.washington.edu/helix

Genetics & Ethics
www.genetics.ca/

Genetics Education for Nurses
www.cincinnatichildrens.org/ed/clinical/gennet/default.htm

Genetics Home Reference
www.ghr.nlm.nih.gov/

Information for Genetic Professionals
www.kumc.edu/gpo/geneinfo.html

International Society of Nurses in Genetics (ISONG)
www.nursing.creighton.edu/isong
References


Online Mendelian Inheritance in Man (OMIM) www.ncbi.nlm.nih.gov/Omim

Organization of Teratogen Information Services www.ots.org

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References


