Chapter 2: Heredity, Prenatal Development, and Birth

In this chapter, we will begin by examining some of the ways in which heredity helps to shape the way we are. We will look at what happens genetically during conception, and describe some known genetic and chromosomal disorders. Next, we will consider what happens during prenatal development, including the impact of teratogens. We will also discuss the impact that both the mother and father have on the developing fetus. Lastly, we will present the birth process and some of the complications that can occur during delivery. Before going into these topics, however, it is important to understand how genes and chromosomes affect development.

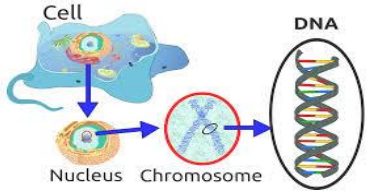
Learning Objectives: Heredity

- ***Define genes***
- ***Distinguish between mitosis and meiosis, genotype and phenotype, homozygous and heterozygous, and dominant and recessive***
- ***Describe some genetic disorders, due to a gene defect, and chromosomal disorders***
- ***Define polygenic and incomplete dominance***
- ***Describe the function of genetic counseling and why individuals may seek genetic counseling***
- ***Define behavioral genetics, describe genotype-environment correlations and genotype-environmental interactions, and define epigenetics***

Heredity

As your recall from chapter one, nature refers to the contribution of genetics to one's development. The basic building block of the nature perspective is the gene. **Genes** *are specific sequence of nucleotides and are recipes for making proteins*. Proteins are responsible for influencing the structure and functions of cells. Genes are located on the chromosomes and there are an estimated 20,500 genes for humans, according to the Human Genome Project (NIH, 2015). See Box 2.2 at the end of this section for more details on the Human Genome Project.

Figure 2.1



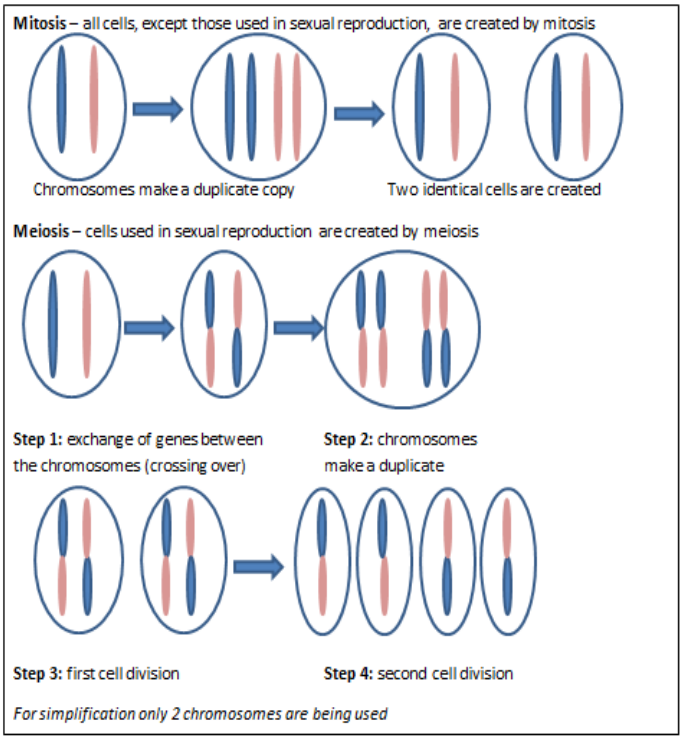
Source

Normal human cells contain 46 chromosomes (or 23 pairs; one from each parent) in the nucleus of the cells. After conception, most cells of the body are created by a process called mitosis. **Mitosis** *is defined as the cell's nucleus making an exact copy of all the chromosomes and splitting into two new cells*. However, the cells used in sexual reproduction, called the gametes (sperm or ova), are formed in a process called meiosis. In **meiosis** *the gamete's chromosomes duplicate, and then divide twice resulting in four cells containing only half the genetic material of the original gamete.* Thus, each sperm and egg possesses only 23 chromosomes and combine to produce the normal 46. See Figure 2.2 for details on both mitosis and meiosis. Given the

amount of genes present and the unpredictability of the meiosis process, the likelihood of having offspring that are genetically identical (and not twins) is one in trillions (Gould & Keeton, 1997).

Of the 23 pairs of chromosomes created at conception, 22 pairs are similar in length. These are called autosomes. The remaining pair, or sex chromosomes, may differ in length. If a child receives the combination of XY the child will be genetically male. If the child receives the combination XX the child will be genetically female.

Figure 2.2 Mitosis vs. Meiosis



Genotypes and Phenotypes

The word **genotype** *refers to the sum total of all the genes a person inherits*. The word **phenotype** *refers to the features that are actually expressed*. Look in the mirror. What do you see, your genotype or your phenotype? What determines whether or not genes are expressed? Because genes are inherited in pairs on the chromosomes, *we may receive either the same version of a gene from our mother and father, that is, be* **homozygous** for that characteristic the gene influences. *If we receive a different version of the gene from each parent, that is referred to as* **heterozygous.** In the homozygous situation we will display that characteristic. It is in the heterozygous condition that it becomes clear that not all genes are created equal. Some genes are **dominant**, meaning they *express themselves in the phenotype even when paired with a different version of the gene,* while their silent partner is called recessive. **Recessive** genes *express themselves only when paired with a similar version gene*. Geneticists refer to *different versions of a gene as* **alleles.** Some dominant traits include having facial dimples, curly hair, normal vision, and dark hair. Some recessive traits include red hair, being nearsighted, and straight hair.

Most characteristics are not the result of a single gene; they are **polygenic**, *meaning they are the result of several genes*. In addition, the dominant and recessive patterns described above are usually not that simple either. *Sometimes the dominant gene does not completely suppress the recessive gene; this is called* **incomplete dominance**. An example of this can be found in the recessive gene disorder sickle cell disease. The gene that produces healthy round-shaped red blood cells is dominant. The recessive gene causes an abnormality in the shape of red blood cells; they take on a sickle form, which can clog the veins and deprive vital organs of oxygen and

increase the risk of stroke. To inherit the disorder a person must receive the recessive gene from both parents. *Those who have inherited only one recessive-gene are called* **carriers** and should be unaffected by this recessive trait. Yet, carriers of sickle cell have some red blood cells that take on the c-shaped sickle pattern. Under circumstances of oxygen deprivation, such as high altitudes or physical exertion, carriers for the sickle cell gene may experience some of the symptoms of sickle cell (Berk, 2004).

Box 2.1 Monozygotic and Dizygotic Twins

Many students are interested in twins. **Monozygotic** *or identical twins occur when a fertilized egg splits apart in the first two weeks of development.* The result is the creation of two separate, but genetically identical offspring. That is, they possess the same genotype and often the same phenotype. About one-third of twins are monozygotic twins. Sometimes, however, *two eggs or ova are released and fertilized by two separate sperm. The result is* **dizygotic** *or fraternal twins*. These two individuals share the same amount of genetic material as would any two children from the same mother and father. In other words, they possess a different genotype and phenotype. Older mothers are more likely to have dizygotic twins than are younger mothers, and couples who use fertility drugs are also more likely to give birth to dizygotic twins. Consequently, there has been an increase in the number of fraternal twins recently (Bortolus et al., 1999).

Source: Monozygotic Twins

Source Dizygotic Twins

Genetic Disorders

Most of the known genetic disorders are dominant gene-linked; however, the vast majority of dominant gene linked disorders are not serious or debilitating. For example, the majority of those with Tourette's Syndrome suffer only minor tics from time to time and can easily control their symptoms. Huntington's Disease is a dominant gene linked disorder that affects the nervous system and is fatal, but does not appear until midlife. Recessive gene disorders, such as cystic fibrosis and sickle-cell anemia, are less common, but may actually claim more lives because they are less likely to be detected as people are unaware that they are carriers of the disease. Some genetic disorders are **sex-linked**; *the defective gene is found on the X-chromosome*. Males have only one X chromosome so are at greater risk for sex-linked disorders due to a recessive gene,

such as hemophilia, color-blindness, and baldness. For females to be affected by the genetic defects, they need to inherit the recessive gene on both X-chromosomes, but if the defective gene is dominant, females can be equally at risk. Table 2.1 lists several genetic disorders.

Table 2.1 Genetic Disorders

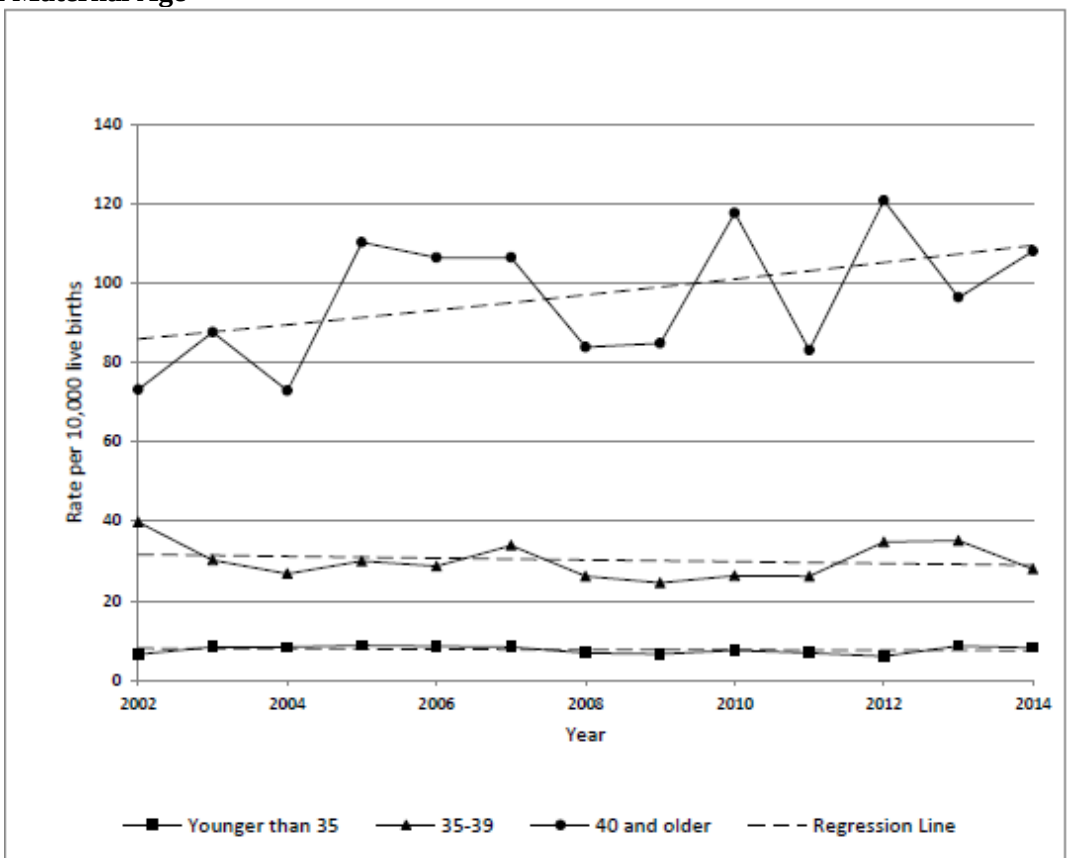
Recessive Disorders (Homozygous): The individual inherits a gene change from both parents. If the gene is inherited from just one parent, the person is a carrier and does not have the condition.	**Cases per Birth**
• **Sickle Cell Disease** (SCD) is a condition in which the red blood cells in the body are shaped like a sickle (like the letter C) and affect the ability of the blood to transport oxygen. Carriers may experience some effects, but do not have the full condition.	1 in 500 Black births 1 in 36,000 Hispanic births
• **Cystic Fibrosis** (CF) is a condition that affects breathing and digestion due to thick mucus building up in the body, especially the lungs and digestive system. In CF, the mucus is thicker than normal and sticky.	1 in 3500
• **Phenylketonuria (PKU)** is a metabolic disorder in which the individual cannot metabolize phenylalanine, an amino acid. Left untreated intellectual deficits occur. PKU is easily detected and is treated with a special diet.	1 in 10,000
• **Tay Sachs Disease** is caused by enzyme deficiency resulting in the accumulation of lipids in the nerve cells of the brain. This accumulation results in progressive damage to the cells and a decrease in cognitive and physical development. Death typically occurs by age five.	1 in 4000 1in 30 American Jews is a carrier 1 in 20 French Canadians is a carrier
• **Albinism** is when the individual lacks melanin and possesses little to no pigment in the skin, hair, and eyes. Vision problems can also occur.	Fewer than 20,000 US cases per year
Autosomal Dominant Disorders (Heterozygous): In order to have the disorder, the individual only needs to inherit the gene change from one parent.	**Cases per Birth**
• **Huntington's Disease** is a condition that affects the individual's nervous system. Nerve cells become damaged, causing various parts of the brain to deteriorate. The disease affects movement, behavior and cognition. It is fatal, and occurs at midlife.	1 in 10,000
• **Tourette Syndrome** is a tic disorder which results in uncontrollable motor and vocal tics as well as body jerking.	1 in 250
• **Achondroplasia** is the most common form of disproportionate short stature. The individual has abnormal bone growth resulting in short stature, disproportionately short arms and legs, short fingers, a large head, and specific facial features.	1 in 15,000-40,000
Sex-Linked Disorders: When the X chromosome carries the mutated gene, the disorder is referred to as an X-linked disorder. Males are more affected than females because they possess only one X chromosome without an additional X chromosome to counter the harmful gene.	**Cases per Birth**
• **Fragile X Syndrome** occurs when the body cannot make enough of a protein it needs for the brain to grow and problems with learning and behavior can occur. Fragile X syndrome is caused from an abnormality in the X chromosome, which then breaks. If a female has fragile X, her second X chromosome usually is healthy, but males with fragile X don't have a second healthy X chromosome. This is why symptoms of fragile X syndrome usually are more serious in males.	1 in 4000 males 1 in 8000 females
• **Hemophilia** occurs when there are problems in blood clotting causing both internal and external bleeding.	1 in 10,000 males
• **Duchenne Muscular Dystrophy** is a weakening of the muscles resulting in an inability to move, wasting away, and possible death.	1 in 3500 males

Chromosomal Abnormalities

A **chromosomal abnormality** *occurs when a child inherits too many or two few chromosomes*. The most common cause of chromosomal abnormalities is the age of the mother. As the mother ages, the ovum is more likely to suffer abnormalities due to longer term exposure to environmental factors. Consequently, some gametes do not divide evenly when they are forming. Therefore, some cells have more than 46 chromosomes. In fact, it is believed that close to half of all zygotes have an odd number of chromosomes. Most of these zygotes fail to develop and are spontaneously aborted by the mother's body.

One of the most common chromosomal abnormalities is on pair 21. **Trisomy 21 or Down syndrome** *occurs when there are three rather than two 21st chromosomes.* A person with Down syndrome typically exhibits an intellectual disability and possesses certain physical features, such as short fingers and toes, folds of skin over the eyes, and a protruding tongue. There is as much variation in people with Down syndrome as in most populations, and those differences need to be recognized and appreciated. Refer to Table 2.2 on the prevalence of Down syndrome in our home state of Illinois. Other less common chromosomal abnormalities of live-born infants occur on chromosome 13 and chromosome 18.

Table 2.2: Illinois Prevalence Rates (2002-2014) for Down Syndrome (Trisomy 21) based on Maternal Age



Source

When the abnormality is on 23rd pair the result is a **sex-linked chromosomal abnormality**. A person might have XXY, XYY, XXX, XO. Two of the more common sex-linked chromosomal disorders are Turner syndrome and Klinefelter syndrome. **Turner syndrome** *occurs when part or all of one of the X chromosomes is lost and the resulting zygote has an XO composition*. This occurs in 1 of every 2,500 live female births (Carroll, 2007) and affects the individual's cognitive functioning and sexual maturation. The external genitalia appear normal, but breasts and ovaries do not develop fully and the woman does not menstruate. Turner syndrome also results in short stature and other physical characteristics. **Klinefelter syndrome (XXY)** *results when an extra X chromosome is present in the cells of a male* and occurs in 1 out of 650 live male births. The Y chromosome stimulates the growth of male genitalia, but the additional X chromosome inhibits this development. An individual with Klinefelter syndrome typically has small testes, some breast development, infertility, and low levels of testosterone (National Institutes of Health, 2019). See Table 2.3 for Chromosomal Disorders descriptions.

Table 2.3 Chromosomal Disorders

Autosomal Chromosome Disorders: The individual inherits too many or two few chromosomes.	**Cases per Birth**
• **Down Syndrome/Trisomy 21** is caused by an extra chromosome 21 and includes a combination of birth defects. Affected individuals have some degree of intellectual disability, characteristic facial features, often heart defects, and other health problems. The severity varies greatly among affected individuals.	1 in 691 1 in 300 births at age 35
• **Trisomy 13** is caused by an extra chromosome 13. Affected individuals have multiple birth defects and generally die in the first weeks or months of life.	1 in 7,906
• **Trisomy 18** is caused by an extra chromosome 18 and the affected individual also has multiple birth defects and early death.	1 in 3,762
Sex-Linked Chromosomal Disorders: The disorder occurs on chromosome pair #23 or the sex chromosomes.	**Cases per Birth**
• **Turner Syndrome** is caused when all or part of one of the X chromosomes is lost before or soon after conception due to a random event. The resulting zygote has an XO composition. Turner Syndrome affects cognitive functioning and sexual maturation in girls. Infertility and a short stature may be noted.	1 in 2500 females
• **Klinefelter Syndrome** is caused when an extra X chromosome is present in the cells of a male due to a random event. The Y chromosome stimulates the growth of male genitalia, but the additional X chromosome inhibits this development. The male can have some breast development, infertility, and low levels of testosterone.	1 in 650 males

Genetic Counseling: *A service that assists individuals identify, test for, and explain potential genetic conditions that could adversely affect themselves or their offspring* is referred to as **genetic counseling** (CDC, 2015b). The common reasons for genetic counseling include:

- Family history of a genetic condition
- Membership in a certain ethnic group with a higher risk of a genetic condition
- Information regarding the results of genetic testing, including blood tests, amniocentesis, or ultra sounds

- Learning about the chances of having a baby with a genetic condition if the parents are older, have had several miscarriages, have offspring with birth defects, experience infertility, or have a medical condition

Behavioral Genetics

Behavioral Genetics *is the scientific study of the interplay between the genetic and environmental contributions to behavior.* Often referred to as the nature/nurture debate, Gottlieb (1998, 2000, 2002) suggests an analytic framework for this debate that recognizes the interplay between the environment, behavior, and genetic expression. This bidirectional interplay suggests that the environment can affect the expression of genes just as genetic predispositions can impact a person's potentials. Additionally, environmental circumstances can trigger symptoms of a genetic disorder. For example, a person who has sickle cell anemia, a recessive gene linked disorder, can experience a sickle cell crisis under conditions of oxygen deprivation. Someone predisposed genetically for type-two diabetes can trigger the disease through poor diet and little exercise.

Research has shown how the environment and genotype interact in several ways. **Genotype-Environment Correlations** *refer to the processes by which genetic factors contribute to variations in the environment* (Plomin et al., 2013). There are three types of genotype-environment correlations:

Figure 2.3

Passive genotype-environment correlation *occurs when children passively inherit the genes and the environments their family provides.* Certain behavioral characteristics, such as being athletically inclined, may run in families. The children have inherited both the genes that would enable success at these activities, and given the environmental encouragement to engage in these actions. Figure 2.3 highlights this correlation by demonstrating how a family passes on water skiing skills through both genetics and environmental opportunities.

Evocative genotype-environment correlation *refers to how the social environment reacts to individuals based on their inherited characteristics.* For example, whether one has a more outgoing or shy temperament will affect how he or she is treated by others.

Active genotype-environment correlation *occurs when individuals seek out environments that support their genetic tendencies*. This is also referred to as *niche picking*. For example, children who are musically inclined seek out music instruction and opportunities that facilitate their natural musical ability.

Conversely, **Genotype-Environment Interactions** *involve genetic susceptibility to the environment.* Adoption studies provide evidence for genotype-environment interactions. For example, the Early Growth and Development Study (Leve et al., 2010) followed 360 adopted children and their adopted and biological parents in a longitudinal study. Results have shown that children whose biological parents exhibited psychopathology, exhibited significantly fewer behavior problems when their adoptive parents used more structured parenting than unstructured. Additionally, elevated psychopathology in adoptive parents increased the risk for the children's development of behavior problems, but only when the biological parents' psychopathology was high. Consequently, the results show how environmental effects on behavior differ based on the genotype, especially stressful environments on genetically at-risk children.

Lastly, **Epigenetics** studies modifications in DNA that affect gene expression and are passed on when the cells divide. Environmental factors, such as nutrition, stress, and teratogens are thought to change gene expression by switching genes on and off. These gene changes can then be inherited by daughter cells. This would explain why monozygotic or identical twins may increasingly differ in gene expression with age. For example, Fraga et al. (2005) found that when examining differences in DNA, a group of monozygotic twins were indistinguishable during the early years. However, when the twins were older there were significant discrepancies in their gene expression, most likely due to different experiences. These differences included susceptibilities to disease and a range of personal characteristics.

Box 2.2 The Human Genome Project

In 1990 the Human Genome Project (HGP), an international scientific endeavor, began the task of sequencing the 3 billion base pairs that make up the human genome. In April of 2003, more than two years ahead of schedule, scientists gave us the genetic blueprint for building a human. Since then, using the information from the HGP, researchers have discovered the genes involved in over 1800 diseases. In 2005 the HGP amassed a large data base called HapMap that catalogs the genetic variations in 11 global populations. Data on genetic variation can improve our understanding of differential risk for disease and reactions to medical treatments, such as drugs. Pharmacogenomic researchers have already developed tests to determine whether a patient will respond favorably to certain drugs used in the treatment of breast cancer, lung cancer or HIV by using information from HapMap (NIH, 2015).

Future directions for the HGP include identifying the genetic markers for all 50 major forms of cancer (The Cancer Genome Atlas), continued use of the HapMap for creating more effective drugs for the treatment of disease, and examining the legal, social and ethical implications of genetic knowledge (NIH, 2015).

From the outset, the HGP made ethical issues one of their main concerns. Part of the HGP's budget supports research and holds workshops that address these concerns. Who owns this information, and how the availability of genetic information may influence healthcare and its impact on individuals, their families, and the greater community are just some of the many questions being addressed (NIH, 2015).

Learning Objectives: Prenatal Development

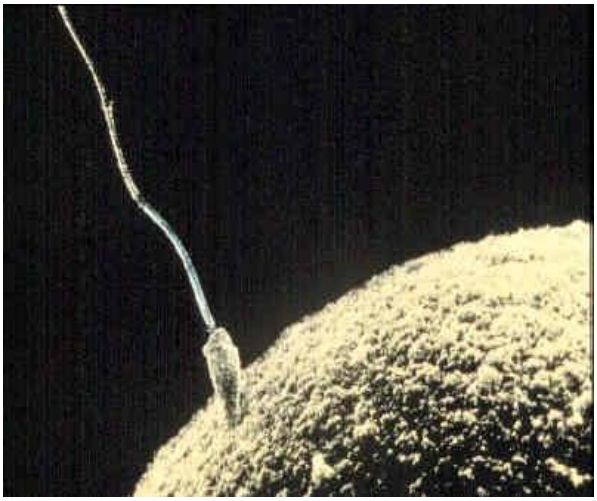
- ***Describe the changes that occur in the three periods of prenatal development***
- ***Describe what occurs during prenatal brain development***
- ***Define teratogens and describe the factors that influence their effects***
- ***List and describe the effects of several common teratogens***
- ***Explain maternal and paternal factors that affect the developing fetus***
- ***Explain the types of prenatal assessment***
- ***Describe both the minor and major complications of pregnancy***

Prenatal Development

Now we turn our attention to prenatal development which is divided into three periods: The germinal period, the embryonic period, and the fetal period. The following is an overview of some of the changes that take place during each period.

The Germinal Period

Figure 2.4



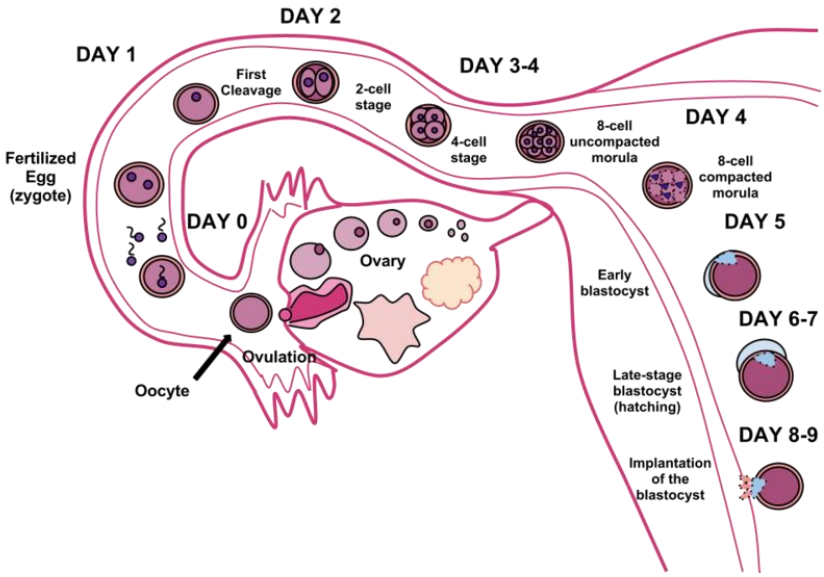
Sperm and Ovum at Conception

The germinal period (about 14 days in length) lasts from conception to implantation of the fertilized egg in the lining of the uterus (See Figure 2.5). At ejaculation millions of sperm are released into the vagina, but only a few reach the egg and typically only one fertilizes the egg. Once a single sperm has entered the wall of the egg, the wall becomes hard and prevents other sperm from entering. After the sperm has entered the egg, the tail of the sperm breaks off and the head of the sperm, containing the genetic information from the father, unites with the nucleus of the egg. It is typically fertilized in the top section of the fallopian tube and continues its journey to the uterus. As a result, a new cell is formed. *This cell, containing the combined genetic information from both parents*, is referred to as a **zygote.**

During this time, the organism begins cell division through mitosis. After five days of mitosis there are 100 cells, which is now called a **blastocyst**. *The blastocyst consists of both an inner and outer group of cells. The inner group of cells, or* **embryonic disk** *will become the embryo*, while *the outer group of cells,* or **trophoblast**, *becomes the support system which nourishes the developing organism*. This stage ends when the blastocyst fully implants into the uterine wall (U.S. National Library of Medicine, 2015a). Approximately 50-75% of blastocysts do not implant in the uterine wall (Betts et al., 2019).

Mitosis is a fragile process and fewer than one half of all zygotes survive beyond the first two weeks (Hall, 2004). Some of the reasons for this include the egg and sperm do not join properly, thus their genetic material does not combine, there is too little or damaged genetic material, the zygote does not replicate, or the blastocyst does not implant into the uterine wall. The failure rate is higher for in vitro conceptions. Figure 2.5 illustrates the journey of the ova from its release to its fertilization, cell duplication, and implantation into the uterine lining.

Figure 2.5 Germinal Period



"Human Fertilization" by Ttrue12 Source

The Embryonic Period

Starting the third week the blastocyst has implanted in the uterine wall. *Upon implantation this multi-cellular organism* is called an **embryo**. Now blood vessels grow forming the **placenta**. *The placenta is a structure connected to the uterus that provides nourishment and oxygen from the mother to the developing embryo via the umbilical cord.* During this period, cells continue to differentiate. Growth during prenatal development occurs in two major directions: *from head to tail* called **cephalocaudal development** and *from the midline outward* referred to as **proximodistal development**. This means that those structures nearest the head develop before those nearest the feet and those structures nearest the torso develop before those away from the

Figure 2.6 The Embryo

Photo by Lunar Caustic

center of the body (such as hands and fingers). The head develops in the fourth week and the precursor to the heart begins to pulse. In the early stages of the embryonic period, gills and a tail are apparent. However, by the end of this stage they disappear and the organism takes on a more human appearance. Some organisms fail during the embryonic period, usually due to gross chromosomal abnormalities. As in the case of the germinal period, often the mother does not yet know that she is pregnant. It is during this stage that the major structures of the body are taking form making the embryonic period the time when the organism is most vulnerable to the greatest amount of damage if exposed to harmful substances. Potential mothers are not often aware of the risks they introduce to the developing embryo during this time. The embryo is approximately 1 inch in length and weighs about 8 grams at the end of eight weeks (Betts et al., 2019). The embryo can move and respond to touch at this time.

The Fetal Period

From the ninth week until birth, the organism is referred to as a **fetus**. During this stage, the major structures are continuing to develop. By the third month, the fetus has all its body parts including external genitalia. In the following weeks, the fetus will develop hair, nails, teeth and the excretory and digestive systems will continue to develop. The fetus is about 3 inches long and weighs about 28 grams.

During the 4th - 6th months, the eyes become more sensitive to light and hearing develops. The respiratory system continues to develop, and reflexes such as sucking, swallowing and hiccupping, develop during the 5th month. Cycles of sleep and wakefulness are present at this time as well. *The first chance of survival outside the womb, known as the* **age of viability** *is reached at about 24 weeks* (Morgan, Goldenberg, & Schulkin, 2008). Many practitioners hesitate to resuscitate before 24 weeks. The majority of the neurons in the brain have developed by 24 weeks, although they are still rudimentary, and the glial or nurse cells that support neurons continue to grow. At 24 weeks the fetus can feel pain (Royal College of Obstetricians and Gynecologists, 1997).

Figure 2.7 Fetus

Source

Between the 7th - 9th months, the fetus is primarily preparing for birth. It is exercising its muscles and its lungs begin to expand and contract. The fetus gains about 5 pounds and 7 inches during this last trimester of pregnancy, and during the 8th month a layer of fat develops under the skin. This layer of fat serves as insulation and helps the baby regulate body temperature after birth.

At around 36 weeks the fetus is almost ready for birth. It weighs about 6 pounds and is about 18.5 inches long. By week 37 all of the fetus's organ systems are developed enough that it could survive outside the mother's uterus without many of the risks associated with premature birth. The fetus continues to gain weight and grow in length until approximately 40 weeks. By then the fetus has very little room to move around and birth becomes imminent. The progression through the stages is shown in Figure 2.8.

Figure 2.8 Prenatal Development Age Milestones

Source

Prenatal Brain Development

Prenatal brain development begins in the third gestational week with the differentiation of stem cells, which are capable of producing all the different cells that make up the brain (Stiles & Jernigan, 2010). *The location of these stem cells in the embryo is referred to as the* **neural plate.** By the end of the third week, two ridges appear along the neural plate first forming the neural groove and then the neural tube. The open region in the center of the neural tube forms the brain's ventricles and spinal canal. By the end of the embryonic period, or week eight, the neural tube has further differentiated into the forebrain, midbrain, and hindbrain.

Brain development during the fetal period involves neuron production, migration, and differentiation. From the early fetal period until midgestation, most of the 85 billion neurons have been generated and many have already migrated to their brain positions. **Neurogenesis,** or *the formation of neurons,* is largely completed after five months of gestation. One exception is in the hippocampus, which continues to develop neurons throughout life. Neurons that form the neocortex, or the layer of cells that lie on the surface of the brain, migrate to their location in an orderly way. Neural migration is mostly completed in the cerebral cortex by 24 weeks (Poduri & Volpe, 2018). Once in position, neurons begin to produce dendrites and axons that begin to form the neural networks responsible for information processing. *Regions of the brain that contain the cell bodies are referred to as the* **gray matter** because they look gray in appearance. *The axons that form the neural pathways make up the* **white matter** because they are covered in myelin, a fatty substance that is white in appearance. Myelin aids in both the insulation and efficiency of neural transmission. Although cell differentiation is complete at birth, the growth of dendrites, axons, and synapses continue for years.

Teratogens

Good prenatal care is essential. The developing child is most at risk for some of the severe problems during the first three months of development. Unfortunately, this is a time at which many mothers are unaware that they are pregnant. Today, we know many of the factors that can jeopardize the health of the developing child. *The study of factors that contribute to birth defects is called* **teratology. Teratogens** *are environmental factors that can contribute to birth defects,* and include some maternal diseases, pollutants, drugs and alcohol.

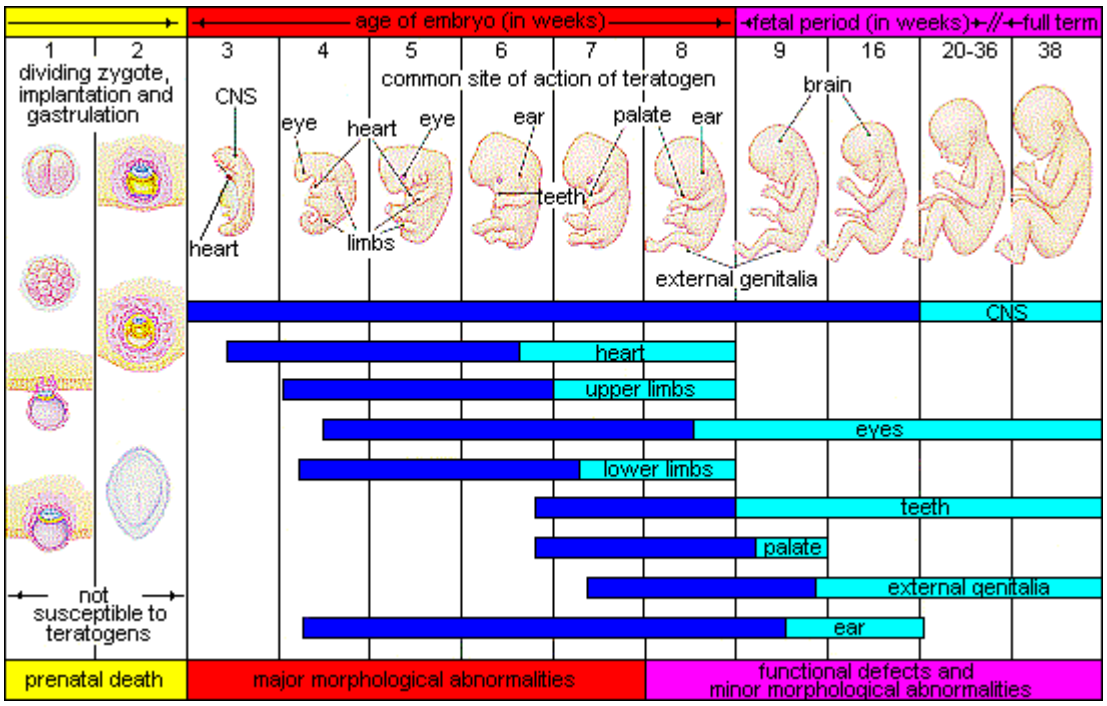
Factors influencing prenatal risks: There are several considerations in determining the type and amount of damage that might result from exposure to a particular teratogen (Berger, 2005). These include:

- **The timing of the exposure**: Structures in the body are vulnerable to the most severe damage when they are forming. If a substance is introduced during a particular structure's critical period (time of development), the damage to that structure may be greater. For example, the ears and arms reach their critical periods at about 6 weeks after conception. If a mother exposes the embryo to certain substances during this period, the arms and ears may be malformed.
- **The amount of exposure**: Some substances are not harmful unless the amounts reach a certain level. The critical level depends in part on the size and metabolism of the mother.

- **The number of teratogens**: Fetuses exposed to multiple teratogens typically have more problems than those exposed to only one.
- **Genetics:** Genetic make-up also plays a role on the impact a particular teratogen might have on the child. This is suggested by fraternal twins exposed to the same prenatal environment, but they do not experience the same teratogenic effects. The genetic make-up of the mother can also have an effect; some mothers may be more resistant to teratogenic effects than others.
- **Being male or female**: Males are more likely to experience damage due to teratogens than are females. It is believed that the Y chromosome, which contains fewer genes than the X, may have an impact.

Figure 2.9 illustrates the timing of teratogen exposure and the types of structural defects that can occur during the prenatal period.

Figure 2.9 Critical Periods of Prenatal Development

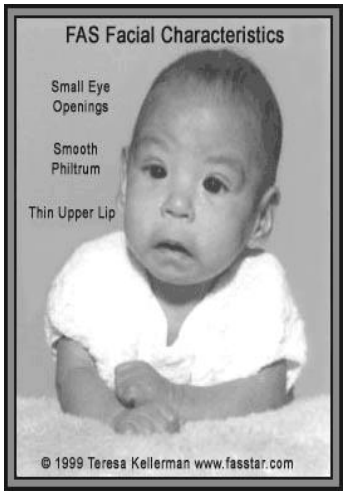


Source

Alcohol: One of the most commonly used teratogens is alcohol, and because half of all pregnancies in the United States are unplanned, it is recommended that women of child-bearing age take great caution against drinking alcohol when not using birth control or when pregnant (CDC, 2005). Alcohol use during pregnancy is the leading preventable cause of intellectual disabilities in children in the United States (Maier & West, 2001). Alcohol consumption, particularly during the second month of prenatal development but at any point during pregnancy, may lead to neurocognitive and behavioral difficulties that can last a lifetime.

In extreme cases, alcohol consumption during pregnancy can lead to fetal death, but also can result in **Fetal Alcohol Spectrum Disorders (FASD),** *which is an umbrella term for the range of effects that can occur due to alcohol consumption during pregnancy* (March of Dimes, 2016a). The most severe form of FASD is Fetal Alcohol Syndrome (FAS). Children with FAS share certain physical features such as flattened noses, small eye holes, and small heads (see Figure 2.10). Cognitively, these children have poor judgment, poor impulse control, higher rates of ADHD, learning issues, and lower IQ scores. These developmental problems and delays persist into adulthood (Streissguth et al., 1996) and can include criminal behavior, psychiatric problems, and unemployment (CDC, 2016a). Based on animal studies, it has been hypothesized that a mother's alcohol consumption during pregnancy may predispose her child to like alcohol (Youngentob et al., 2007). **Binge drinking**, *or 4 or more drinks in 2 to 3 hours,* during pregnancy increases the chance of having a baby with FASD (March of Dimes, 2016a).

Figure 2.10 Fetal Alcohol Spectrum Disorders



Fetal Alcohol Spectrum Disorder Facial Features

Facial Feature	Potential Effect of Fetal Alcohol
Head size	Below-average head circumference
Eyes	Smaller than average eye opening, skin folds at corner of eyes
Nose	Low nasal bridge, short nose
Midface	Smaller than average midface size
Lip and philtrum	Thin upper lip, indistinct philtrum

Source

Tobacco: Another widely used teratogen is tobacco as more than 7% of pregnant women smoked in 2016 (Someji & Beltrán-Sánchez, 2019). According to Tong et al. (2013) in conjunction with the Centers for Disease Control and Prevention, data from 27 sites in 2010 representing 52% of live births, showed that among women with recent live births:

- About 23% reported smoking in the 3 months prior to pregnancy.
- Almost 11% reported smoking during pregnancy.
- More than half (54.3%) reported that they quit smoking by the last 3 months of pregnancy.
- Almost 16% reported smoking after delivery.

When comparing the ages of women who smoked:

- Women <20, 13.6% smoked during pregnancy
- Women 20–24,17.6% smoked during pregnancy
- Women 25–34, 8.8% smoked during pregnancy
- Women ≥35, 5.7% smoked during pregnancy

The findings among racial and ethnic groups indicated that smoking during pregnancy was highest among American Indians/Alaska Natives (26.0%) and lowest among Asians/Pacific Islanders (2.1%).

When a pregnant woman smokes the fetus is exposed to dangerous chemicals including nicotine, carbon monoxide and tar, which lessen the amount of oxygen available to the fetus. Oxygen is important for overall growth and development. Tobacco use during pregnancy has been associated with low birth weight, **ecotopic pregnancy** (*fertilized egg implants itself outside of the uterus*), **placenta previa** (*placenta lies low in the uterus and covers all or part of the cervix*), **placenta abruption** (*placenta separates prematurely from the uterine wall*), preterm delivery, stillbirth, fetal growth restriction, sudden infant death syndrome (SIDS), birth defects, learning disabilities, and early puberty in girls (Center for Disease Control, 2015d).

A woman being exposed to secondhand smoke during pregnancy has also been linked to low-birth weight infants. In addition, exposure to **thirdhand smoke**, *or toxins from tobacco smoke that linger on clothing, furniture, and in locations where smoking has occurred,* results in a negative impact on infants' lung development. Rehan et al., (2011) found that prenatal exposure to thirdhand smoke played a greater role in altered lung functioning in children than exposure postnatally.

Prescription/Over-the-counter Drugs: About 70% of pregnant women take at least one prescription drug (March of Dimes, 2016e). A woman should not be taking any prescription drug during pregnancy unless it was prescribed by a health care provider who knows she is pregnant. Some prescription drugs can cause birth defects, problems in overall health, and development of the fetus. Over-the-counter drugs are also a concern during the prenatal period because they may cause certain health problems. For example, the pain reliever ibuprofen can cause serious blood flow problems to the fetus during the last three months.

Figure 2.11

Source

Illicit Drugs: Common illicit drugs include cocaine, ecstasy and other club drugs, heroin, marijuana, and prescription drugs that are abused. It is difficult to completely determine the effects of a particular illicit drug on a developing child because most mothers who use, use more than one substance and have other unhealthy behaviors. These include smoking, drinking alcohol, not eating healthy meals, and being more likely to get a sexually transmitted disease. However, several problems seem clear. The use of cocaine is connected with low birth weight, stillbirths and spontaneous abortion. Heavy marijuana use is associated with problems in brain development (March of Dimes, 2016c). *If a baby's mother used an addictive drug during pregnancy that baby can*

get addicted to the drug before birth and go through drug withdrawal after birth, also known as **Neonatal abstinence syndrome** (March of Dimes, 2015d). Other complications of illicit drug use include premature birth, smaller than normal head size, birth defects, heart defects, and infections. Additionally, babies born to mothers who use drugs may have problems later in life, including learning and behavior difficulties, slower than normal growth, and die from sudden infant death syndrome. Children of substance abusing parents are also considered at high risk for a range of biological, developmental, academic, and behavioral problems, including developing substance abuse problems of their own (Conners et al., 2003).

Box 2.3 Should Women Who Use Drugs During Pregnancy Be Arrested and Jailed?

Women who use drugs or alcohol during pregnancy can cause serious lifelong harm to their child. Some people have advocated mandatory screenings for women who are pregnant and have a history of drug abuse, and if the women continue using, to arrest, prosecute, and incarcerate them (Figdor & Kaeser, 1998). This policy was tried in Charleston, South Carolina 20 years ago. The policy was called the Interagency Policy on Management of Substance Abuse During Pregnancy and had disastrous results:

> The Interagency Policy applied to patients attending the obstetrics clinic at MUSC, which primarily serves patients who are indigent or on Medicaid. It did not apply to private obstetrical patients. The policy required patient education about the harmful effects of substance abuse during pregnancy. A statement also warned patients that protection of unborn and newborn children from the harms of illegal drug abuse could involve the Charleston police, the Solicitor of the Ninth Judicial Court, and the Protective Services Division of the Department of Social Services (DSS). (Jos et al., 1995, pp. 120–121)

This policy seemed to deter women from seeking prenatal care, deterred them from seeking other social services, and was applied solely to low-income women, resulting in lawsuits. The program was canceled after 5 years, during which 42 women were arrested. A federal agency later determined that the program involved human experimentation without the approval and oversight of an institutional review board (IRB).

In July 2014, Tennessee enacted a law that allows women who illegally use a narcotic drug while pregnant to be prosecuted for assault if her infant is harmed or addicted to the drug (National Public Radio, 2015). According to the National Public Radio report, a baby is born dependent on a drug every 30 minutes in Tennessee, which is a rate three times higher than the national average. However, since the law took effect the number of babies born having drug withdrawal symptoms has not diminished. Critics contend that the criminal justice system should not be involved in what is considered a healthcare problem. What do you think? Is the issue of mothers using illicit drugs more of a legal or medical concern?

Pollutants: There are more than 83,000 chemicals used in the United States with little information on the effects of them during pregnancy (March of Dimes, 2016b).

- **Lead:** An environmental pollutant of significant concern is lead poisoning, which has been linked to fertility problems, high blood pressure, low birth weight, prematurity, miscarriage, and slowed neurological development. Grossman and Slutsky (2017) found that babies born in Flint Michigan, an area identified with high lead levels in the drinking water, were premature, weighed less than average, and gained less weight than expected.
- **Pesticides:** The chemicals in certain pesticides are also potentially damaging and may lead to birth defects, learning problems, low birth weight, miscarriage, and premature birth (March of Dimes, 2014).
- **Bisphenol A**: Prenatal exposure to bisphenol A (BPA), a chemical commonly used in plastics and food and beverage containers, may disrupt the action of certain genes contributing to certain birth defects (March of Dimes, 2016b).
- **Radiation:** If a mother is exposed to radiation, it can get into the bloodstream and pass through the umbilical cord to the baby. Radiation can also build up in body areas close to the uterus, such as the bladder. Exposure to radiation can slow the baby's growth, cause birth defects, affect brain development, cause cancer, and result in a miscarriage.
- **Mercury:** Mecury, a heavy metal, can cause brain damage and affect the baby's hearing and vision. This is why women are cautioned about the amount and type of fish they consume during pregnancy.

Toxoplasmosis: *The tiny parasite, toxoplasma gondii, causes an infection called* **toxoplasmosis**. According to the March of Dimes (2012d), toxoplasma gondii infects more than 60 million people in the United States. A healthy immune system can keep the parasite at bay producing no symptoms, so most people do not know they are infected. As a routine prenatal screening frequently does not test for the presence of this parasite, pregnant women may want to talk to their health-care provider about being tested. Toxoplasmosis can cause premature birth, stillbirth, and can result in birth defects to the eyes and brain. While most babies born with this infection show no symptoms, ten percent may experience eye infections, enlarged liver and spleen, jaundice, and pneumonia. To avoid being infected, women should avoid eating undercooked or raw meat and unwashed fruits and vegetables, touching cooking utensils that touched raw meat or unwashed fruits and vegetables, and touching cat feces, soil or sand. If women think they may have been infected during pregnancy, they should have their baby tested.

Figure 2.12

Source

Sexually Transmitted Diseases: Gonorrhea, syphilis, and chlamydia are sexually transmitted infections that can be passed to the fetus by an infected mother. Mothers should be tested as early as possible to minimize the risk of spreading these infections to their unborn child. Additionally, the earlier the treatment begins, the better the health outcomes for mother and baby (CDC, 2016d). Sexually transmitted diseases (STDs) can cause premature birth, premature rupture of

the amniotic sac, an ectopic pregnancy, birth defects, miscarriage, and still births (March of Dimes, 2013). Most babies become infected with STDS while passing through the birth canal during delivery, but some STDs can cross the placenta and infect the developing fetus.

Human Immunodeficiency Virus (HIV): One of the most potentially devastating teratogens is HIV. HIV and Acquired Immune Deficiency Syndrome (AIDS) are leading causes of illness and death in the United States (Health Resources and Services Administration, 2015). One of the main ways children under age 13 become infected with HIV is via mother-to-child transmission of the virus prenatally, during labor, or by breastfeeding (CDC, 2016c). There are some measures that can be taken to lower the chance the child will contract the disease. HIV positive mothers who take antiviral medications during their pregnancy greatly reduce the chance of passing the virus to the fetus. The risk of transmission is less than 2 percent; in contrast, it is 25 percent if the mother does not take antiretroviral drugs (CDC, 2016b). However, the long-term risks of prenatal exposure to the medication are not known. It is recommended that women with HIV deliver the child by c-section, and that after birth they avoid breast feeding.

German measles (or rubella): Rubella, also called German measles, is an infection that causes mild flu-like symptoms and a rash on the skin. However, only about half of children infected have these symptoms, while others have no symptoms (March of Dimes, 2012a). Rubella has been associated with a number of birth defects. If the mother contracts the disease during the first three months of pregnancy, damage can occur in the eyes, ears, heart or brain of the unborn child. Deafness is almost certain if the mother has German measles before the 11th week of prenatal development and can also cause brain damage. Women in the United States are much less likely to be afflicted with rubella, because most women received childhood vaccinations that protect her from the disease.

Maternal Factors

Mothers over 35: Most women over 35 who become pregnant are in good health and have healthy pregnancies. However, according to the March of Dimes (2016d), women over age 35 are more likely to have an increased risk of:

- Fertility problems
- High blood pressure
- Diabetes
- Miscarriages
- Placenta Previa
- Cesarean section
- Premature birth
- Stillbirth
- A baby with a genetic disorder or other birth defects

Because a woman is born with all her eggs, environmental teratogens can affect the quality of the eggs as women get older. Also, a woman's reproductive system ages which can adversely affect the pregnancy. Some women over 35 choose special prenatal screening tests, such as a maternal blood screening, to determine if there are any health risks for the baby.

Figure 2.13

Source

Although there are medical concerns associated with having a child later in life, there are also many positive consequences to being a more mature parent. Older parents are more confident, less stressed, and typically married providing family stability. Their children perform better on math and reading tests, and they are less prone to injuries or emotional troubles (Albert, 2013). Women who choose to wait are often well educated and lead healthy lives. According to Gregory (2007), older women are more stable, demonstrate a stronger family focus, possess greater self-confidence, and have more money. Having a child later in one's career equals overall higher wages. In fact, for every year a woman delays motherhood, she makes 9% more in lifetime earnings. Lastly, women who delay having children actually live longer. Sun et al. (2015) found that women who had their last child after the age of 33 doubled their chances of living to age 95 or older than women who had their last child before their 30th birthday. A woman's natural ability to have a child at a later age indicates that her reproductive system is aging slowly, and consequently so is the rest of her body.

Teenage Pregnancy: A teenage mother is at a greater risk for having pregnancy complications including anemia, and high blood pressure. These risks are even greater for those under age 15. Infants born to teenage mothers have a higher risk for being premature and having low birthweight or other serious health problems. Premature and low birthweight babies may have organs that are not fully developed which can result in breathing problems, bleeding in the brain, vision loss, and serious intestinal problems. Very low birthweight babies (less than 3 1/3 pounds) are more than 100 times as likely to die, and moderately low birthweight babies (between 3 1/3 and 5 ½ pounds) are more than 5 times as likely to die in their first year, than normal weight babies (March of Dimes, 2012c). Again, the risk is highest for babies of mothers under age 15. Reasons for these health issues include that teenagers are the least likely of all age groups to get early and regular prenatal care. Additionally, they may engage in negative behaviors including eating unhealthy food, smoking, drinking alcohol, and taking drugs. Additional concerns for teenagers are repeat births. About 25% of teen mothers under age 18 have a second baby within 2 years after the first baby's birth.

Gestational Diabetes: Seven percent of pregnant women develop **gestational diabetes** (March of Dimes, 2015b). *Diabetes is a condition where the body has too much glucose in the bloodstream*. Most pregnant women have their glucose level tested at 24 to 28 weeks of pregnancy. Gestational diabetes usually goes away after the mother gives birth, but it might indicate a risk for developing diabetes later in life. If untreated, gestational diabetes can cause premature birth, stillbirth, the baby having breathing problems at birth, jaundice, or low blood sugar. Babies born to mothers with gestational diabetes can also be considerably heavier (more than 9 pounds) making the labor and birth process more difficult. For expectant mothers, untreated gestational diabetes can cause preeclampsia (high blood pressure and signs that the liver and kidneys may not be working properly) discussed later in the chapter. Risk factors for gestational diabetes include age (being over age 25), being overweight or gaining too much

weight during pregnancy, family history of diabetes, having had gestational diabetes with a prior pregnancy, and race and ethnicity (African-American, Native American, Hispanic, Asian, or Pacific Islander have a higher risk). Eating healthy and maintaining a healthy weight during pregnancy can reduce the chance of gestational diabetes. Women who already have diabetes and become pregnant need to attend all their prenatal care visits, and follow the same advice as those for women with gestational diabetes as the risk of preeclampsia, premature birth, birth defects, and stillbirth are the same.

High Blood Pressure (Hypertension): Hypertension *is a condition in which the pressure against the wall of the arteries becomes too high.* There are two types of high blood pressure during pregnancy, gestational and chronic. Gestational hypertension only occurs during pregnancy and goes away after birth. Chronic high blood pressure refers to women who already had hypertension before the pregnancy or to those who developed it during pregnancy and it continued after birth. According to the March of Dimes (2015c) about 8 in every 100 pregnant women have high blood pressure. High blood pressure during pregnancy can cause premature birth and low birth weight (under five and a half pounds), placental abruption, and mothers can develop preeclampsia.

Rh Disease: Rh is a protein found in the blood. Most people are Rh positive, meaning they have this protein. Some people are Rh negative, meaning this protein is absent. Mothers who are Rh negative are at risk of having a baby with *a form of anemia* called **Rh disease** (March of Dimes, 2009). A father who is Rh-positive and mother who is Rh-negative can conceive a baby who is Rh-positive. Some of the fetus's blood cells may get into the mother's bloodstream and her immune system is unable to recognize the Rh factor. The immune system starts to produce antibodies to fight off what it thinks is a foreign invader. Once her body produces immunity, the antibodies can cross the placenta and start to destroy the red blood cells of the developing fetus. As this process takes time, often the first Rh positive baby is not harmed, but as the mother's body will continue to produce antibodies to the Rh factor across her lifetime, subsequent pregnancies can pose greater risk for an Rh positive baby. In the newborn, Rh disease can lead to jaundice, anemia, heart failure, brain damage and death.

Weight Gain during Pregnancy: According to March of Dimes (2016f) during pregnancy most women need only an additional 300 calories per day to aid in the growth of the fetus. Gaining too little or too much weight during pregnancy can be harmful. Women who gain too little may

Table 2.4 Weight Gain during Pregnancy

If you were a healthy weight before pregnancy	If you were underweight before pregnancy	If you were overweight before pregnancy	If you were obese before pregnancy
• gain 25-35lbs • 1-4½lbs in the first trimester and 1lb per week in the second and third trimesters	• gain 28-40lbs • 1-4½lbs in the first trimester and a little more than 1lb per week thereafter	• gain 12-25 lbs • 1-4½lbs in the first trimester and a little more than ½lb per week in the second and third trimesters	• 11-20lbs • 1-4½lbs in the first trimester and less than ½lb per week in the second and third trimesters
Mothers of twins need to gain more in each category.			

Source

have a baby who is low-birth weight, while those who gain too much are likely to have a premature or large baby. There is also a greater risk for the mother developing preeclampsia and diabetes, which can cause further problems during the pregnancy. Table 2.4 shows the healthy weight gain during pregnancy. Putting on the weight slowly is best. Mothers who are concerned about their weight gain should talk to their health care provider.

Stress: Feeling stressed is common during pregnancy, but high levels of stress can cause complications including having a premature baby or a low-birthweight baby. Babies born early or too small are at an increased risk for health problems. Stress-related hormones may cause these complications by affecting a woman's immune systems resulting in an infection and premature birth. Additionally, some women deal with stress by smoking, drinking alcohol, or taking drugs, which can lead to problems in the pregnancy. High levels of stress in pregnancy have also been correlated with problems in the baby's brain development and immune system functioning, as well as childhood problems such as trouble paying attention and being afraid (March of Dimes, 2012b).

Depression: Depression is a significant medical condition in which feelings of sadness, worthlessness, guilt, and fatigue interfere with one's daily functioning. Depression can occur before, during, or after pregnancy, and 1 in 7 women is treated for depression sometime between the year before pregnancy and year after pregnancy (March of Dimes, 2015a). Women who have experienced depression previously are more likely to have depression during pregnancy. Consequences of depression include the baby being born premature, having a low birthweight, being more irritable, less active, less attentive, and having fewer facial expressions. About 13% of pregnant women take an antidepressant during pregnancy. It is important that women taking antidepressants during pregnancy discuss the medication with a health care provider as some medications can cause harm to the developing organism. In fact, birth defects happen about 2 to 3 times more often in women who are prescribed certain Selective Serotonin Reuptake Inhibitors (SSRIs) for their depression.

Paternal Impact: The age of fathers at the time of conception is also an important factor in health risks for children. According to Nippoldt (2015) offspring of men over 40 face an increased risk of miscarriages, autism, birth defects, achondroplasia (bone growth disorder) and schizophrenia. These increased health risks are thought to be due to accumulated chromosomal aberrations and mutations during the maturation of sperm cells in older men (Bray et al., 2006). However, like older women, the overall risks are small.

Figure 2.14 Hazardous Occupations

Source

In addition, men are more likely than women to work in occupations where hazardous chemicals, many of which have teratogenic effects or may cause genetic mutations, are used (Cordier, 2008). These may include petrochemicals, lead, and pesticides that can cause abnormal sperm and lead to miscarriages or diseases. Men are also more likely to be a source of secondhand smoke for their developing offspring. As noted earlier, smoking by either the mother or around the mother can hinder prenatal development.

Prenatal Assessment

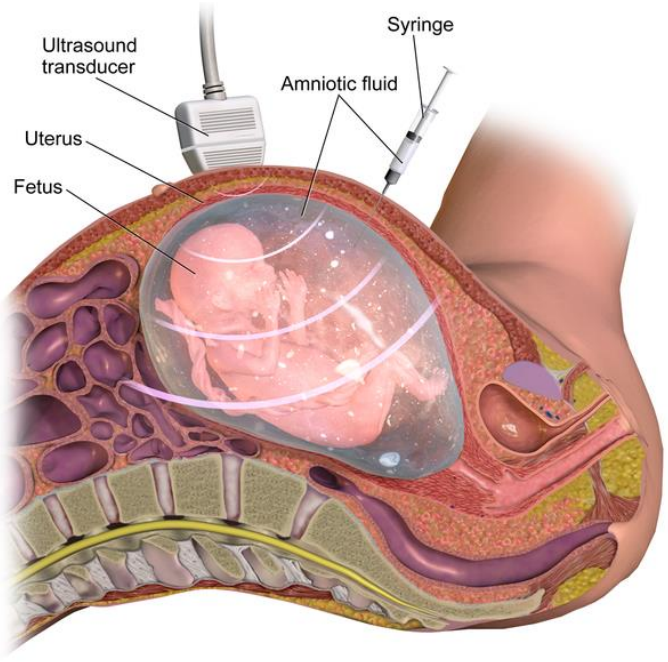
A number of assessments are suggested to women as part of their routine prenatal care to find conditions that may increase the risk of complications for the mother and fetus (Eisenberg et al., 1996). These can include blood and urine analyses and screening and diagnostic tests for birth defects.

Figure 2.15 Preparing for an Ultrasound

Source

Ultrasound is one of the main screening tests done in combination with blood tests. The ultrasound *is a test in which sound waves are used to examine the fetus*. There are two general types. Transvaginal ultrasounds are used in early pregnancy, while transabdominal ultrasounds are more common and used after 10 weeks of pregnancy (typically, 16 to 20 weeks). Ultrasounds are used to check the fetus for defects or problems. It can also find out the age of the fetus, location of the placenta, fetal position, movement, breathing and heart rate, amount of amniotic fluid, and number of fetuses. Most women have at least one ultra sound during pregnancy, but if problems are noted, additional ultrasounds may be recommended.

Figure 2.16 Amniocentesis



Source

When diagnosis of a birth defect is necessary, ultrasounds help guide the more invasive diagnostic tests of amniocentesis and chorionic villus sampling. **Amniocentesis** *is a procedure in which a needle is used to withdraw a small amount of amniotic fluid and cells from the sac surrounding the fetus and later tested* (see Figure 2.16).

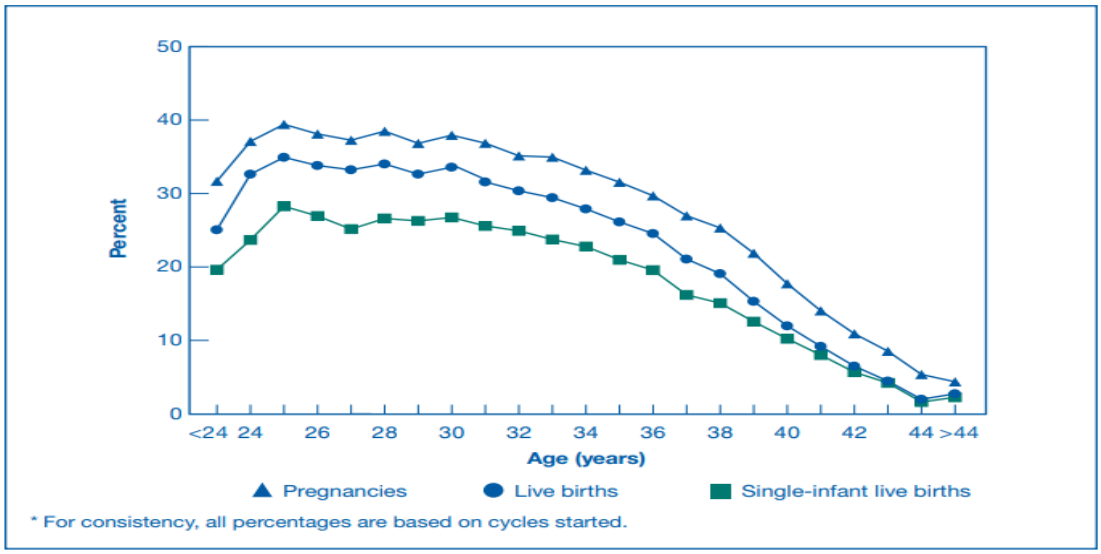
Chorionic Villus Sampling *is a procedure in which a small sample of cells is taken from the placenta and tested.* Both amniocentesis and chorionic villus sampling have a risk of miscarriage, and consequently they are not done routinely.

Box 2.4 Infertility and Reproductive Technology

Infertility: Infertility affects about 10 to 15 percent of couples in the United States (Mayo Clinic, 2015). For men, the most common cause is a lack of, or low sperm production, and for women, it is the failure to ovulate. Another common cause for women is p**elvic inflammatory disease** (**PID**), *which is an infection of a woman's reproductive organs* (Carroll, 2007). It is often a complication caused by and STD, such as chlamydia and gonorrhea, although other infections that are not sexually transmitted can also cause PID.

Fertility treatment: The majority of infertility cases are treated using fertility drugs to increase ovulation, or with surgical procedures to repair the reproductive organs or remove scar tissue from the reproductive tract. In **in vitro fertilization (IVF)** *eggs are removed from the female and are fertilized outside the woman's body*. The fertilized egg is then reinserted in the woman's uterus. The success rate varies depending on the type of egg implanted, such as whether the egg was recently removed from the woman, used after being frozen, or donated from another woman. Success is also highly dependent on the age of the mother (See Figure 2.17).

Figure 2.17 Percentage of Pregnancies, Live Births, and Single-Infant Live Births from IVF from Fresh Non-Donor Eggs



Source

Higher success rates, but less common procedures include **gamete intra-fallopian tube transfer** (GIFT) *which involves implanting both sperm and ova into the fallopian tube and fertilization is allowed to occur naturally* (Carroll, 2007). **Zygote intra-fallopian tube transfer** (ZIFT) is *another procedure in which sperm and ova are fertilized outside of the woman's body and the fertilized egg is then implanted in the fallopian tube*. This allows the zygote to travel down the fallopian tube and embed in the lining of the uterus naturally. This procedure also has a higher success rate than IVF.

Complications of Pregnancy

Minor complications: There are a number of common side effects of pregnancy. Not everyone experiences all of these, nor to the same degree. And although they are considered "minor" this is not to say that these problems are not potentially very uncomfortable. These side effects include nausea (particularly during the first 3-4 months of pregnancy as a result of higher levels of estrogen in the system), heartburn, gas, hemorrhoids, backache, leg cramps, insomnia, constipation, shortness of breath or varicose veins (as a result of carrying a heavy load on the abdomen).

Major Complications: The following are some serious complications of pregnancy which can pose health risks to mother and child and that often require hospitalization.

Ectopic Pregnancy *occurs when the zygote becomes attached to the fallopian tube before reaching the uterus.* About 1 in 50 pregnancies in the United States are tubal pregnancies and this number has been increasing because of the higher rates of pelvic inflammatory disease and Chlamydia (Carroll, 2007). Abdominal pain, vaginal bleeding, nausea and fainting are symptoms of ectopic pregnancy.

Preeclampsia, also known as Toxemia, *is characterized by a sharp rise in blood pressure, a leakage of protein into the urine as a result of kidney problems, and swelling of the hands, feet, and face during the third trimester of pregnancy*. Preeclampsia is the most common complication of pregnancy. It is estimated to affect 5% to 10% of all pregnancies globally and accounts for 40% to 60% of maternal deaths in developing countries (National Institute of Child Health and Human Development, 2013). Rates are lower in the United States and preeclampsia affects about 3% to 5% of pregnant women.

Preeclampsia occurs most frequently in first pregnancies, and it is more common in women who are obese, have diabetes, or are carrying twins. *When preeclampsia causes seizures, the condition is known as* **eclampsia**, which is the second leading cause of maternal death in the United States. Preeclampsia is also a leading cause of fetal complications, which include low birth weight, premature birth, and stillbirth. Treatment is typically bed rest and sometimes medication. If this treatment is ineffective, labor may be induced.

Maternal Mortality: Acording to the CDC (2019), about 700 American women die from complications related to pregnancy each year, and this number is rising. Further, 60% of those deaths could have been prevented. Bleeding, infections, and heart-related problems are the main causes. Possible contributing factors include the high caesarean section rate and obesity. Compared to other developed nations, this number is considered high. Approximately 1000 women die in childbirth around the world each day (World Health Organization, 2010). Rates are highest in Subsaharan Africa and South Asia, although there has been a substantial decrease in these rates. The campaign to make childbirth safe for everyone has led to the development of clinics accessible to those living in more isolated areas and training more midwives to assist in childbirth.

Spontaneous abortion is experienced in an estimated 20-40 percent of undiagnosed pregnancies and in another 10 percent of diagnosed pregnancies. Usually the body aborts due to chromosomal abnormalities, and this typically happens before the 12th week of pregnancy. Cramping and bleeding result and normal periods return after several months. Some women are more likely to have repeated miscarriages due to chromosomal, amniotic, or hormonal problems, but miscarriage can also be a result of defective sperm (Carrell et. al., 2003).

Learning Objectives: Birth

- ***Describe how expectant parents prepare for childbirth***
- ***Describe the stages of vaginal delivery***
- ***Explain why a caesarean or induced birth is necessary***
- ***Describe the two common procedures to assess the condition of the newborn***
- ***Describe problems newborns experience before, during, and after birth***

Preparation for Childbirth

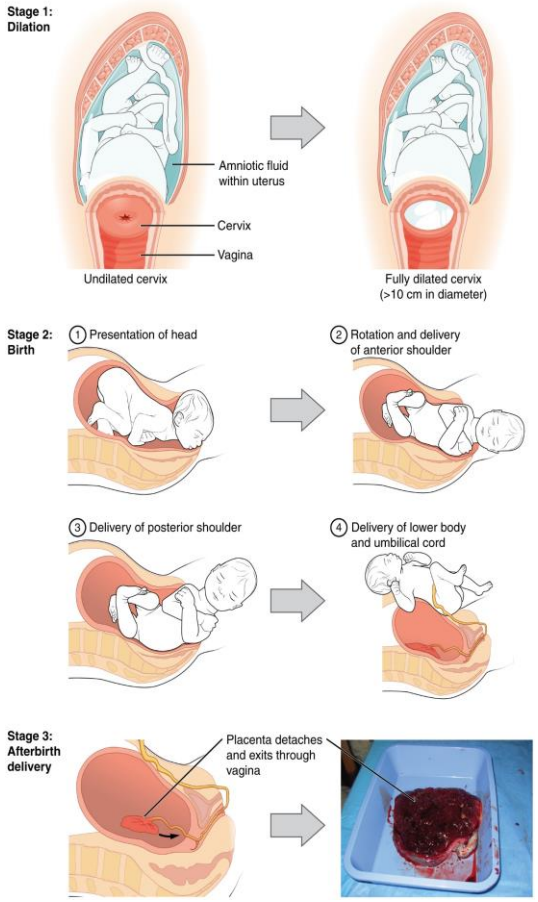
Prepared childbirth refers to being not only in good physical condition to help provide a healthy environment for the baby to develop, but also helping individuals to prepare to accept their new roles as parents. Additionally, parents can receive information and training that will assist them for delivery and life with the baby. The more future parents can learn about childbirth and the newborn, the better prepared they will be for the adjustment they must make to a new life.

One of the most common methods for preparing for childbirth is **The Lamaze Method.** This method originated in Russia and was brought to the United States in the 1950s by Fernand Lamaze. *The emphasis of this method is on teaching the woman to be in control in the process of delivery.* It includes learning muscle relaxation, breathing though contractions, having a focal point (usually a picture to look at) during contractions and having a support person who goes through the training process with the mother and serves as a coach during delivery (Eisenberg et al., 1996).

Choosing Where to Have the Baby and Who Will Deliver: The vast majority of births occur in a hospital setting. However, one percent of women choose to deliver at home (Martin et al., 2015). Women who are at low risk for birth complications can successfully deliver at home. More than half (67%) of home deliveries are by certified nurse midwifes. Midwives are trained and licensed to assist in delivery and are far less expensive than the cost of a hospital delivery. However, because of the potential for a complication during the birth process, most medical professionals recommend that delivery take place in a hospital. Despite the concerns, in the United States women who have had previous children, who are over 25, and who are white are more likely to have out-of-hospital births (MacDorman et al., 2010). In addition to home births, one-third of out-of-hospital births occur in freestanding clinics, birthing centers, in physician's offices, or other locations.

Stages of Birth for Vaginal Delivery

Figure 2.17 Stages of Birth for a Vaginal Delivery



Source

The First Stage of labor begins with uterine contractions that may initially last about 30 seconds and be spaced 15 to 20 minutes apart. These increase in duration and frequency to more than a minute in length and about 3 to 4 minutes apart. Typically, doctors advise that they be called when contractions are coming about every 5 minutes. Some women experience *false labor or* **Braxton-Hicks contractions**, especially with the first child. These may come and go. They tend to diminish when the mother begins walking around. Real labor pains tend to increase with walking. Labor may also be signaled by a bloody discharge being expelled from the cervix. In one out of 8 pregnancies, the amniotic sac or water in which the fetus is suspended may break before labor begins. In such cases, the physician may induce labor with the use of medication if it does not begin on its own in order to reduce the risk of infection. Normally this sac does not rupture until the later stages of labor.

The first stage of labor is typically the longest. During this stage the cervix or opening to the uterus dilates to 10 centimeters or just under 4 inches (See Figure 2.17). This may take around 12-16 hours for first children or about 6-9 hours for women who have previously given birth. Labor may also begin with a discharge of blood or amniotic fluid.

The Second Stage involves the passage of the baby through the birth canal. This stage takes about 10-40 minutes. Contractions usually come about every 2-3 minutes. The mother pushes and relaxes as directed by the medical staff. Normally the head is delivered first. The baby is then rotated so that one shoulder can come through and then the other shoulder. The rest of the baby quickly passes through. At this stage, an **episiotomy** *or incision made in the tissue between the vaginal opening and anus*, may be performed to avoid tearing the tissue of the back of the vaginal opening (Mayo Clinic, 2016). The baby's mouth and nose are suctioned out. The umbilical cord is clamped and cut.

The Third Stage is relatively painless. During this stage, the placenta or afterbirth is delivered. This is typically within 20 minutes after delivery. If an episiotomy was performed it is stitched up during this stage.

More than 50% of women giving birth at hospitals use an epidural anesthesia during delivery (American Pregnancy Association, 2015). An **epidural block** *is a regional analgesic that can be used during labor and alleviates most pain in the lower body without slowing labor.* The epidural block can be used throughout labor and has little to no effect on the baby. Medication is injected into a small space outside the spinal cord in the lower back. It takes 10 to 20 minutes for the medication to take effect. An epidural block with stronger medications, such as anesthetics, can be used shortly before a C-section or if a vaginal birth requires the use of forceps or vacuum extraction.

A **Cesarean section (C-section)** *is surgery to deliver the baby by being removed through the mother's abdomen.* In the United States, about one in three women have their babies delivered this way (Martin et al., 2015). Most C-sections are done when problems occur during delivery unexpectedly. These can include:

- Health problems in the mother
- Signs of distress in the baby
- Not enough room for the baby to go through the vagina
- The position of the baby, such as a breech presentation where the head is not in the downward position

C-sections are also more common among women carrying more than one baby. Although the surgery is relatively safe for mother and baby, it is considered major surgery and carries health risks. Additionally, it also takes longer to recover from a C-section than from vaginal birth. After healing, the incision may leave a weak spot in the wall of the uterus. This could cause problems with an attempted vaginal birth later. However, more than half of women who have a C-section can have a vaginal birth later.

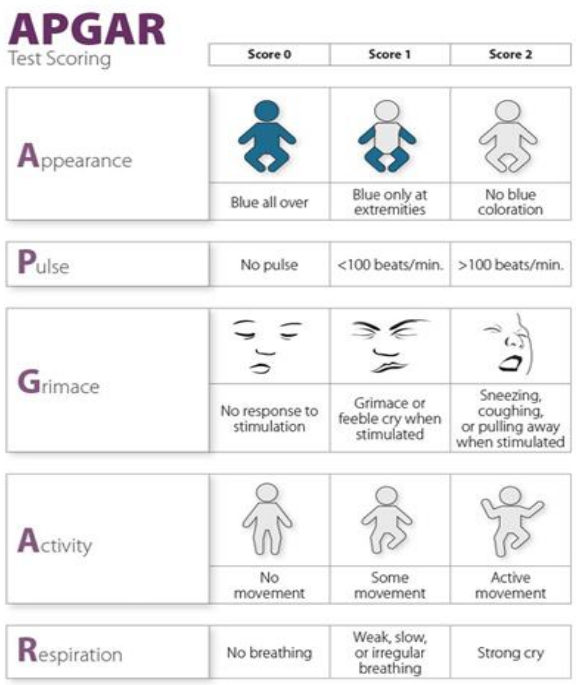
Induced birth: Sometimes a baby's arrival may need to be **induced** *or delivered before labor begins*. Inducing labor may be recommended for a variety of reasons when there is concern for the health of the mother or baby. For example:

- The mother is approaching two weeks beyond her due date and labor has not started naturally
- The mother's water has broken, but contractions have not begun
- There is an infection in the mother's uterus
- The baby has stopped growing at the expected pace
- There is not enough amniotic fluid surrounding the baby
- The placenta peels away, either partially or completely, from the inner wall of the uterus before delivery
- The mother has a medical condition that might put her or her baby at risk, such as high blood pressure or diabetes (Mayo Clinic, 2014)

Assessing the Neonate

The Apgar assessment is conducted one minute and five minutes after birth. This is a very quick way to assess the newborn's overall condition. Five measures are assessed: Heart rate, respiration, muscle tone (assessed by touching the baby's palm), reflex response (the Babinski reflex is tested), and color. A score of 0 to 2 is given on each feature examined. An Apgar of 5 or less is cause for concern. The second Apgar should indicate improvement with a higher score (see Figure 2.18).

Figure 2.18 APGAR Scores



Source:

Another way to assess the condition of the newborn is the Neonatal Behavioral Assessment Scale (NBAS). The baby's motor development, muscle tone, and stress response are assessed. This tool has been used around the world to further assess the newborn, especially those with low Apgar scores, and to make comparisons of infants in different cultures (Brazelton & Nugent, 1995).

Problems of the Newborn

Figure 2.19 Newborn in Neonatal Unit

Source

Anoxia: **Anoxia** *is a temporary lack of oxygen to the brain*. Difficulty during delivery may lead to anoxia which can result in brain damage or in severe cases, death. Babies who suffer both low birth weight and anoxia are more likely to suffer learning disabilities later in life as well.

Low Birth weight: We have been discussing a number of teratogens associated with low birth weight such as alcohol, tobacco, etc. A child is considered **low birth weight** *if he or she weighs less than 5 pounds 8 ounces (2500 grams).* About 8.2 percent of babies born in the United States are of low birth weight (CDC, 2015a). A low birth weight baby has difficulty maintaining adequate body temperature

because it lacks the fat that would otherwise provide insulation. Such a baby is also at more risk for infection, and 67 percent of these babies are also preterm which can make them more at risk for respiratory infection. Very low birth weight babies (2 pounds or less) have an increased risk of developing cerebral palsy.

Additionally, Pettersson et al., (2019) analyzed fetal growth and found that reduced birth weight was correlated with a small, but significant increase in several psychiatric disorders in adulthood. These included: attention-deficit/hyperactivity disorder, autism, depression, and obsessive-compulsive disorder. Pettersson et al. theorized that "reduced fetal growth compromises brain development during a critical period, which in turn slightly increases the risk not only for neurodevelopmental disorders but also for virtually all mental health conditions" (p. 540). An insufficient supply of oxygen and nutrients for the developing fetus are proposed as factors that increased the risk for neurodevelopmental disorders.

Preterm: A newborn might also have a low birth weight if it is *born at less than 37 weeks gestation*, which qualifies it as a **preterm** baby (CDC, 2015c). Early birth can be triggered by anything that disrupts the mother's system. For instance, vaginal infections can lead to premature birth because such infection causes the mother to release anti-inflammatory chemicals which, in turn, can trigger contractions. Smoking and the use of other teratogens can lead to preterm birth. The earlier a woman quits smoking, the lower the chance that the baby will be born preterm (Someji & Beltrán-Sánchez, 2019). A significant consequence of preterm birth includes **respiratory distress syndrome**, *which is characterized by weak and irregular breathing* (United States National Library of Medicine, 2015b).

Figure 2.20 Saybie

Source

Saybie (name given to her by the hospital), a baby girl born in San Diego, California is now considered the world's smallest baby ever to survive (Chiu, 2019). She was born in December 2018 at 23 weeks and 3 days weighing only 8.6 ounces (same size as an apple). After five months in the hospital, Saybie went home in May 2019 weighing 5 pounds.

Small-for-Date Infants: *Infants that have birth weights that are below expectation based on their gestational age are referred to as* **small-for-date.** These infants may be full term or preterm, but still weigh less than 90 % of all babies of the same gestational age. This is a very serious situation for newborns as their growth was adversely affected. Regev et al. (2003) found that small-for-date infants died at rates more than four times higher than other infants. Remember that many causes of low birth weight and preterm births are preventable with proper prenatal care.

Postpartum Maternal Concerns

After pregnancy many women experience emotional changes. The "baby blues" are often mentioned as a common occurrence in new mothers. The **baby blues** *are feelings of sadness that occur 3 to 5 days after having a baby, and typically disappear usually within 10 days of the birth*. New mothers may have trouble sleeping, be moody, and feel let-down from the birthing experience. However, postpartum depression is not the same as the baby blues. According to the Diagnostic and Statistical Manual of Mental Disorders (5th edition, text revision) (DSM-5-TR), (American Psychiatric Association, 2022), **peripartum onset of depression,** also known as postpartum depression, *is a type of depression that occurs during pregnancy or in the 4 weeks following pregnancy*. Approximately 1 out of 8 women experience postpartum depression and symptoms can include feelings of sadness, sleeplessness, and difficulty bonding with the newborn.

Changing hormone levels are thought to be a factor in the occurrence of peripartum depression, however, risk factors include having depression previously, a family history of depression, being younger than 20, experiencing stress, and substance use. Peripartum-onset mood disorders, both depression and mania, can present with or without psychotic features. Hallucinations and delusions are associated with postpartum psychotic episodes and have included command hallucinations to kill the infant or delusions that the infant is possessed. Psychotic features occur in approximately 1 in 500 to 1 in 1,000 deliveries, and the risk is higher for women with prior postpartum mood episodes (American Psychiatric Association, 2022).

Postpartum anxiety is also a concern for many new mothers. According to Bregel (2017) because oxytocin, a bonding hormone, rises during preganancy, brain areas related to empathy and anxiety are heightened. Consequently, the new mother is "hard-wired" to respond to and fend for her baby, which can lead to toxic levels of stress and anxiety. These can manifest as heightened alertness, intrusive and horrifying thoughts of something terrible happening to the infant, and physiological arousal. Just as for peripartum depression and postpartum psychosis, a new mother experiencing postpartum anxiety should seek assistance from a health care provider.

COVID-19: Impact on Prenatal Development

Vaccines: COVID-19 infections are more severe in pregnant women compared to their nonpregnant peers resulting in greater hospital admissions, intensive care unit stays, and death (Gray et al., 2021). As of March 1, 2021 more than 80 maternal deaths and over 73,600 COVID-19 infections occurred in pregnant women in the United States. Despite these numbers, pregnant women were excluded from the initial COVID-19 vaccine trials. It was not until February 2021 that the first vaccine trials in pregnant women began. Gray et al. reviewed the results of the COVID-19 vaccine on 131 reproductive age women, including 84 who were pregnant, 31 lactating, and 16 nonpregnant. Results indicated that the COVID-19 messenger RNA vaccines generated immunity in pregnant and lactating women similar to that observed in nonpregnant women. Additionally, immunity transferred to the neonates via placenta and breastmilk. Mithal et al. (2021) reported that pregnant women who were vaccinated for COVID-19 earlier in their third trimester had a higher likelihood of passing protective antibodies to their newborns than women who received their vaccine closer to delivery.

Additionally, Shanes et al. (2021) reviewed the findings of 84 women who received a COVID-19 vaccine during pregnancy and 116 women in a control group who did not receive a vaccine. Vaccinated women showed a strong antibody response, and these COVID-19 antibodies were successfully transferred to their fetuses There was also no evidence that the vaccines affected the women's placentas. In contrast, the placentas of 16 women who tested positive for COVID-19 while pregnant showed evidence of injury (Samuelson, 2020). According to pathological exams completed directly following birth, injuries included reduced blood flow from the mother to the fetus due to abnormal blood vessels and blood clots in the placenta. Shanes et al. (2021) concluded that research supports the safety of COVID-19 vaccination in pregnant women without adverse effects for the fetus.

Fetal Alcohol Spectrum Disorder (FASD): During the COVID-19 pandemic, alcohol use increased sharply, and there is evidence that alcohol use among pregnant women also increased (Weir, 2022). Researchers estimate that 2%-5% of U.S. children may be affected by alcohol exposure resulting in behavioral, learning, and mental health disorders. As of July 2022 a bipartisan bill, called the FASD Respect Act, is before Congress. This bill would support FASD research, surveillance, and activities related to diagnosis, prevention and treatment.

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