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Genetics, Birth Disorders and Pregnancy

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Unit Plan This curriculum unit is designed to be used in an alternative public school setting for pregnant teens. It has been planned for general science special education classes. The program includes grades seven through twelve, with students ranging in age from thirteen to nineteen. The students are identified as needing special education, for a variety of reasons. Ability levels cover a wide range. Most students in these classes, however, have limited reading and writing skills. Some students exhibit behaviors which are best addressed in small groups within a highly structured setting. The curriculum unit is planned for students who have little background in genetics or human reproduction. It would also be suitable for use in parenting classes or as a unit in a general biology class or human physiology class in regular education. Individual sections of the unit may be used in isolation based on interest and need.

Goal Students will gain information and develop a better understanding of genetics as it relates to human development, birth defects, and pregnancy. This will be accomplished through the use of selected videos, reading materials, small group discussions and various experiential, hands-on learning activities.

Rational Statistics show that teen pregnancy in the United States continues to increase. This is of particular concern because teen mothers and their babies face increased risks to their health. They need to be informed and educated about pregnancy and related issues. Approximately one million teenagers become pregnant each year, and more than 530,000 give birth. Pregnant teens are the least likely of all maternal age groups to get early and regular prenatal care. A teenage mother is more at risk of pregnancy complications such as premature labor, anemia and high blood pressure. Three million teens are affected by sexually transmitted diseases annually. These include chlamydia, syphilis (which can cause blindness and death to an infant) and AIDS, which is fatal to the mother and can infect the infant. A baby born to a teenage mother is more at risk than one born to an older mother. Nine percent of teenage girls have low-birthweight babies compared to seven percent of all mothers nationally. These babies may have organs that are not fully developed, which can lead to lung problems or bleeding in the brain. Low-birthweight babies are forty times more likely to die in their first month of life than normal-weight babies. Teens need to be informed and educated. They need to understand the risks involved in negative behaviors. Often they have poor eating habits, may smoke, drink alcohol and take drugs. These behaviors greatly increase the risk that their babies will be born with health problems.

Modern genetics is a relatively new field; unfamiliar to many teens. In the past few decades, knowledge has grown in the field of human genetics and there are new discoveries being made daily. Many medical problems

can now be treated or prevented in babies, including various errors of body chemistry. Genes involved in causing high blood cholesterol, muscular dystrophies and cystic fibrosis have been found and are being studied. Identifying these genes is the first step toward developing specific treatment, such as gene therapy. Teens need to be informed and educated about these advances.

BACKGROUND

History Gregor Mendel is often called the father of genetics. In the mid-1800s this young monk worked in the monastery gardens experimenting with pea plants. Mendel began his experiments by studying the stem length of pea plants. He noticed differences. Because pea plants grow and reproduce quickly, Mendel was able to observe many generations. From his experiments, Mendel concluded that traits were passed from the parent plant to their offspring. He hypothesized that certain factors must produce certain traits. Today, Mendel's factors are called genes. We now know that an organism receives two genes for each trait. One gene comes from the female reproductive cell and the second one comes from the male. The stronger of the two genes is called the dominant gene and the gene that is weaker is called the recessive gene. The dominant gene is the gene that always shows itself. The recessive gene is the gene that is hidden when the dominant gene is present. Mendel observed that the tall gene is dominant in pea plants and the short gene is recessive. That is why plants with one tall gene and one short gene are always tall. The results of Mendel's experiments were published in 1866. However, it was not until 1900 that his research was analyzed by scientists. Many new discoveries have been made, and yet, Mendel's original hypothesis still forms the basis of modern genetics.

Modern Genetics Francis Crick and James Watson became the founding fathers of the new molecular genetics by describing the complex structure of DNA in 1953. Other scientists, building on the knowledge of DNA's structure, have `cracked' the genetic code and have begun to identify and locate specific genes. Scientists, working on the Human Genome Project, now hope to map all of the estimated 50,000 to 100,000 human genes and spell out the entire message conveyed by three billion chemical code letters in the human genome. That message, written in the code of DNA, is found in the nucleus of the body's ten trillion cells (except red blood cells). Its instructions not only determine the structure, size, coloring, and other physical attributes of each human being, but can also affect intelligence, susceptibility to disease, and behavior. Commenting on the impact of the Human Genome Project, James Watson has said, "We used to think that our fate was in the stars. Now we know that, in large measure, our fate is in our genes."

Cells The human body is made up of tiny units called cells. Body cells are microscopic in size, and each cell must perform certain functions in order for the whole body to function. Each cell grows and develops by using the food that is brought to it by the blood. Each cell can dispose of its wastes, respond to heat and light, and reproduce. The adult human body may contain a million billion cells. These cells perform different functions and may look different, but they are all alike in basic structure. The main parts of the cells are the *cell membrane, nucleus,* and the liquid *cytoplasm* between these two. The cell membrane controls substances passing in and out of the cell, the nucleus directs the activity of the cell by containing chromosomes, and the cytoplasm provides a medium for the transportation of substances. The basic living substance of all cells is called *protoplasm*

Chromosomes These are threadlike structures in the nucleus of a cell that control heredity. During cell division, each chromosome makes a copy of itself. A pair of identical chromosomes is formed. Each new

daughter cell receives one chromosome from each of the pairs. In both asexual and sexual reproduction, chromosomes are passed from parent to offspring. During asexual reproduction, which occurs in plants, bacteria, and lower organisms, each daughter cell receives its chromosomes from a single parent. During sexual reproduction, the daughter cell receives chromosomes from each parent cell. In this case new organisms contain chromosomes from both of its parents.

Genes These are parts of a chromosome that control inherited traits. Each gene affects a different trait. Genes determine height, eye color, hair color, and many other characteristics. Each gene is located at a certain place on the chromosome. Genes also control the life processes of cells.

DNA This is the chemical that makes up chromosomes. It stands for deoxyribonucleic acid. A molecule of DNA looks like a twisted ladder. The sides of the DNA ladder are made up of sugars and phosphates. The steps of the DNA ladder are made up of four kinds of nitrogen bases. A British scientist, Rosalind Franklin took x-ray photographs of DNA crystals. Based on these photographs, Watson, Crick and Wilkins predicted the shape and composition of the DNA molecule. A single DNA molecule, or ladder, can have thousands of steps. The number and arrangement of these steps form a genetic "code." This code determines the kind of protein that is made. Different genes determine different kinds of inherited traits. During cell division, each chromosome doubles to form a pair of identical chromosomes. Molecules of DNA in the parent chromosome also double. This process by which DNA is duplicated is called replication. The DNA ladder breaks apart between the nitrogen bases in the steps. This is similar to the process of unzipping a zipper. The other nitrogen bases attach to each half of the ladder. The result is two new DNA ladders that are exact copies of the original DNA molecule. Sometimes a DNA molecule does not replicate itself exactly. The chromosomes do not pair correctly. These changes in chromosomes and genes are called mutations. Many mutations are harmful to the organism. However, many also are neutral or occur in non-transcribed regions.

Simplified Concept DNA makes proteins, proteins perform many different functions in all types of cells, and cells make up the organism

DISORDERS

Spina Bifida This is a birth defect of the backbone and sometimes the spinal cord. It is one of a group of birth defects called neural tube defects. The neural tube is the embryonic structure that develops into the brain and spine. Spina bifida is among the most common severe birth defects in the country. Approximately 2,000 babies (one in every 2,000 live births) are born with this condition each year. Spina bifida occurs more frequently in certain racial/ethnic groups, particularly whites of European extraction. It is less common among Jews, Asians, and blacks. Causes of spina bifida are not well understood. Scientists believe that genetic and environmental factors act together to cause this and other neural tube defects. Although spina bifida appears to run in certain families, it does not follow any particular law of inheritance. If parents have one child with spina bifida, the risk of recurrence in any subsequent pregnancy is about one in 40. Studies show that the development of spina bifida and other neural tube defects may be influenced by the mother's diet, especially the amount of the B-vitamin folic acid she consumes. The most important time to take folic acid is at least one month before conception and during the first month of pregnancy, since this is when neural tube defects occur. Spina bifida can often be detected before birth. The AFP screening test detects pregnancies at higher-than-average risk of spina bifida and other neural tube defects. A detailed ultrasound examination to inspect the fetal spine may follow high AFP test results. It has been suggested that caesarean delivery prior to the

onset of labor may reduce the severity of paralysis in babies with spina bifida, although, this is still controversial.

Down Syndrome This is a combination of birth defects including some degree of mental retardation and characteristic facial features. About 30 to 50 percent of babies with Down syndrome also have congenital heart defects, and many have visual and hearing impairment and other health problems. The severity of these problems varies greatly. Down syndrome is one of the most common genetic birth defects. It affects all races and economic levels equally. Approximately one in 800 to one in 1,000 babies are born with the disorder. In this country, there are about 250,000 individuals with Down syndrome. The major cause of Down syndrome is an extra chromosome number 21. The features of Down syndrome result from having this extra chromosome 21 in each of the body's cells. This is called trisomy 21, because of the presence of three number 21 chromosomes. Translocation Down syndrome occurs when the extra chromosome 21 is attached to another chromosome in the egg or sperm. This condition, in either parent, increases the chances of having another child with Down syndrome. The risk of Down syndrome increases with age (1 in 106 at age 40), however, 70 percent of babies with Down syndrome are born to women who are under age 35. Prenatal testing using amniocentesis or CVS can diagnose Down syndrome.

Tay-Sachs Disease This disease is an inherited birth defect. The symptoms first appear at about 6 months, when an apparently normal baby gradually stops smiling, crawling, or turning over, and eventually becomes blind, paralyzed, and unaware of his surroundings. Death occurs by age five. Babies with Tay-Sachs disease lack an enzyme called hex A that is necessary for breaking down certain fatty substances in brain and nerve cells. These fatty substances build up and gradually destroy brain and nerve cells, until the entire central nervous system stops working. Other forms of the disease are referred to as juvenile, chronic and adult-onset forms of hex A deficiency. Both infantile and adult forms of Tay-Sachs disease occur most frequently in descendants of Central and Eastern European Jews. Non-Jewish individuals of French-Canadian ancestry are at similarly increased risk. Both groups have about 100 times the rate of occurrence of other ethnic groups. A Tay-Sachs carrier has one normal gene for hex A and one Tay-Sachs gene. When two carriers become parents there is a one-in-four chance that any child they have will inherit a Tay-Sachs gene from each parent and have the disease. Prenatal amniocentesis and CVS can diagnose Tay-Sachs disease before birth. Generally, if prenatal testing shows that hex A is present, the baby will not have Tay-Sachs disease. If it is missing, the baby will be affected.

Sickle Cell Disease This is an inherited blood disease which can cause bouts of pain, damage to vital organs, and for some, death in childhood or early adulthood. Sickle cell disease affects a protein inside the red blood cells. It occurs when a person inherits two sickle cell genes or a combination of one sickle cell gene plus another abnormal genes that affect the red blood cells. Red blood cells are normally round and flexible. But when oxygen is released by the red blood cells in people with sickle cell disease, the cells become distorted, forming a rigid banana or sickle shape that can clog blood vessels. Sickle cells tend to become trapped and destroyed in the liver and spleen. This results in anemia, a shortage of red blood cells. The effects of sickle cell disease vary greatly from one person to the next. Some affected people rarely see their doctors; others may be hospitalized frequently. To inherit the disease, a child must receive two sickle cell genes- one from each parent who carries the sickle cell gene. In the United States, most cases of sickle cell disease occur among blacks and Hispanics. About one in every 400 to 600 blacks inherits the sickle cell disease. A blood test can identify people who have either sickle cell trait or the disease. There is also a prenatal test to determine whether the fetus will have sickle cell disease, carry the trait, or be unaffected. Infants have an increased chance for fatal infection unless treated with penicillin.

PKU Phenylketonuria is an inherited disorder of body chemistry that, if untreated, causes mental retardation. Most affected newborns are now diagnosed and treated early, allowing them to grow up free of mental retardation. About one baby in 12,000 is born with PKU in the United States. The disorder occurs in all ethnic groups, but it is most common in individuals of Northern European ancestry. PKU is a disease that affects the way the body processes food. Children with PKU cannot process phenylalanine, a constituent of protein. As a result, phenylalanine builds up in the bloodstream and causes brain damage and mental retardation. PKU is inherited when both parents have the PKU gene and pass it on to their baby. When both parents are carriers, there is a one-in-four chance that each will pass the PKU gene on to a child, causing it to be born with the disease. Using a test developed in the 1960s, babies are now tested for PKU before they leave the hospital. The test is highly accurate when performed between 24 hours of age and less than 7 days of age. A second testing at two weeks is recommended in some cases. Mental retardation can be prevented, in affected newborns, if they are treated with a special diet that is low in phenylalanine beginning before the fourth week of life. Individuals with PKU should remain on a restricted diet throughout childhood, and most likely for life.

Marfan Syndrome This syndrome, which affects about one in 10,000 Americans, is one of the most common inherited disorders of connective tissue. It affects males and females from all racial and ethnic groups. Symptoms of Marfan syndrome may be mild or severe, and may be present at birth or appear in adult life. The disorder sometimes causes sudden death in adults who were unaware that they had it. It is one of more than 100 inherited disorders of connective tissue with abnormalities that may affect the heart, blood vessels, lungs, eyes, bones and ligaments. Affected individuals are often tall, slender, and loose-jointed. Arms and legs may be unusually long. The spine may have a curve, and the breastbone may protrude or look caved in. The face may be long and narrow, with a high roof of the mouth and crowded teeth. Heart and blood vessels nearly always are affected. Sudden large splits in the aorta can result in death. Persons with Marfan syndrome are also prone to sudden lung collapse. Individuals are plagued with various eye problems as well. Marfan syndrome is caused by a defective gene on chromosome 15. A whole variety of mutations in this gene can cause the syndrome. Normally, this gene tells the body to produce a protein called fibrillin. Individuals with Marfan syndrome have scant or faulty fibrillin in their affected tissues. The abnormal gene is usually inherited from one parent who has the disorder. It is a dominant gene; each child has a 50-50 chance of inheriting it. There is currently no single, conclusive test to diagnose Marfan syndrome.

Congenital Heart Defects More than 25,000 infants (one out of every 125-150) are born with heart defects each year in the United States. Heart defects are among the most common birth defects, and are the leading cause of birth defect-related deaths. A condition is called congenital when it is present at birth. Heart defects begin in the early part of pregnancy when the heart is forming. Congenital heart defects can affect any of the different parts or functions of the heart. In most cases, scientists do not know what makes a baby’s heart develop abnormally. Both genetic and environmental factors appear to play some role. Women who contract rubella during the first three months of pregnancy have a high risk of having a baby with a heart defect. Other viruses also may play a role in heart defects. Certain medications also increase the risk. These include the acne medication Accutane, lithium, and possibly certain anti-seizure medications, as well as other problems. Drinking alcohol in pregnancy also can increase the risk of heart defects. Studies also suggest that use of cocaine in pregnancy increases the risk of these birth defects. A special form of ultrasound called echo cardiography can accurately detect many heart defects. If certain heart problems are diagnosed before birth, medications often help the problem before the fetal heart starts to fail. In other cases, knowing that a problem exists, enables doctors to be ready to give the baby the treatment it needs as soon as it is born.

Achondroplasia This is a genetic disorder of bone growth that is evident at birth. It affects about one in every 26,000 births and it occurs in all races and in both sexes. It is one of the oldest recorded birth defects. During

fetal development and childhood cartilage normally develops into bone. In individuals with achondroplasia, something goes wrong during this process, especially in the long bones. Only a small amount of cartilage in the growth plates of the long bones turns into bone, leading to short bones and reduced height. Generally, the head is large, and the nose is flat at the bridge. Teeth may be crowded and poorly aligned. Because of the large head, short arms and legs and loose joints, a baby with achondroplasia is slow to sit, stand and walk alone. Most children with Achondroplasia have normal intelligence. Psychological problems may arise because of the difficulties in adjusting to a world geared to normal-sized people. Achondroplasia is caused by an abnormal gene located on one of the chromosome 4 pair. In some cases, a child inherits achondroplasia from a parent who has the condition. In most cases (over 80 percent) achondroplasia is not inherited but results from a new mutation that occurred in the egg or sperm cell that formed the embryo. The parents of children with achondroplasia resulting from new mutations are usually average-sized. Geneticists have observed that older-than-average fathers (aged 50 and older) are somewhat more likely to have children with achondroplasia and certain other autosomal dominant conditions caused by new mutations. Because researchers have identified the gene that causes achondroplasia, highly accurate prenatal tests are available which can diagnose or rule out achondroplasia.

Thalassemia This disorder consists of a group of inherited diseases of the blood. About 100,000 babies worldwide are born with severe forms of the disease each year. It occurs most frequently in people of Italian, Greek, Middle Eastern, Southern Asian and African ancestry. Thalassemia includes a number of different forms of anemia. The most severe form results in fetal or newborn death. Most individuals have milder forms of the disease, with varying degrees of anemia. Many children appear healthy at birth, but become ill during the first year or two of life. They grow slowly and often develop jaundice. Without treatment, the spleen, liver, and heart soon become greatly enlarged. Bones become thin and brittle; face bones become distorted, and children with thalassemia often look alike. Frequent blood transfusions and antibiotics are used to treat the disease when needed. All forms of thalassemia are transmitted only through heredity. The disease is passed on through parents who carry the thalassemia gene in their cells. Blood tests and family genetic studies can show whether an individual has thalassemia or is a carrier. Prenatal testing using CVS or amniocentesis can detect or rule out thalassemia in the fetus. Early diagnosis is important so that treatment can prevent as many complications as possible.

Cleft Lip and Palate A cleft is an opening in the lip, the roof of the mouth, or the soft tissue in the back of the mouth. These openings are normally present in early fetal development, and usually close by the tenth to twelfth week of pregnancy. They fail to close in approximately one in every 700 babies born. Clefts occur more often among Asians and certain groups of American Indians than among whites. They occur less frequently among blacks. The causes of cleft lip and palate are not well understood. Information suggests that they may be the result of a combination of genetic elements with environmental factors, such as drugs, infections, maternal illnesses, and possibly deficiency of folic acid. Children with clefts have special problems, particularly with feeding, ear diseases and speech development, as well as dental problems. Little is known about how to prevent clefts. Studies have shown that fetuses with a certain predisposing gene may be at increased risk of developing cleft palate if their mothers smoke. Other factors, such as maternal alcohol abuse, and maternal diabetes have been linked to increased risk of clefts.

Clubfoot This is one of the most common of all birth defects, affecting some 9,000 babies (about one in 400) born in the United States each year. Boys are affected twice as often as girls. Clubfoot is a term used for several kinds of ankle and foot deformities usually present at birth. The defect can be mild or severe, and it can happen to one foot or both. The exact cause of clubfoot still is not clear in many cases. Many scientists think the defect starts early in pregnancy, before the baby is large enough to stay in one position very long.

Clubfoot is probably caused by a combination of heredity and other factors that may affect prenatal growth. Although the effects of clubfoot often may be prevented through early treatment, there is no method of preventing the defect at this time. Genetic counseling can help parents understand the odds with each pregnancy for having a child with clubfoot.

Cystic Fibrosis This is one of the most common inherited diseases, affecting about one in 2500 Caucasians in the United States. The condition is less common in people of other ethnic or racial backgrounds. Cystic fibrosis results from having two CF genes. Individuals with cystic fibrosis have inherited one CF gene from each parent. A parent who has one CF gene and one normal gene is a carrier. A carrier is not affected with the disease, but may pass on either the CF gene or the normal gene to his offspring. With each pregnancy, there is a one in four chance that the child will inherit two CF genes and will have cystic fibrosis. There is a 75% chance that the child will not have the disease. The offspring could be a carrier (50%), or could inherit two normal genes (25%). These chances are the same for each pregnancy. Cystic fibrosis causes the body to produce large amounts of abnormally thick mucus. This collects in the lungs, causing illness. The pancreas is also often damaged by the disease. Deficiency in pancreas enzymes may cause diarrhea and poor growth Cystic fibrosis does not affect intellect. Currently there is no known cure, however, individuals with the disease are living longer than in the past, often into their late 20's or 30's. Genetic testing is available to determine whether an individual carries the CF gene. This test can find approximately 90% of Caucasian carriers. The rate of accuracy is higher for non-caucasians. In every population some CF carriers will be missed by the test. Prenatal diagnosis involves testing cell samples obtained through CVS or amniocentesis. The prenatal test can tell with great accuracy whether the unborn baby does or does not have genes which result in cystic fibrosis. However, in some instances the results are inconclusive.

Muscular Dystrophy There are several forms of muscular dystrophy, the most common and severe type is called Duchenne Muscular Dystrophy. This is a muscle-wasting disorder which affects boys almost exclusively. The onset is usually between the ages of two and four and progresses rapidly. Few individuals survive their early twenties. There is no known cure at this time. Flaws in muscle protein genes cause muscular dystrophies. These disorders are generally inherited, but in some cases no family history of the disease exists. DNA testing can be done to diagnose or rule out Duchenne or Becker muscular dystrophies. Scientists have discovered the gene that, when defective, is responsible for these dystrophies. Carriers of Duchenne and Becker muscular dystrophies may be identified through protein and DNA-based detection tests.

Hemophilia This is a hereditary disorder in which one of the plasma proteins needed to form a clot is missing or reduced. The most common type of hemophilia is factor VIII deficiency, or hemophilia A. The second most common type is factor IX deficiency or hemophilia B. When a person with hemophilia is injured he will have prolonged bleeding because he cannot make a firm clot. Some bleeding episodes occur as a result of injury, but many occur seemingly without cause. Approximately one in 10,000 males born in the United States has hemophilia. All races are affected equally. Hemophilia is a sex-linked hereditary bleeding disorder transmitted on a gene of the X chromosome. If all of a person's X chromosomes have the hemophilia gene, then that person will have hemophilia. In some cases hemophilia is dormant for many generations if no affected male children are born. The gene for hemophilia in such cases is carried through several generations of females who, because they have a second X chromosome that is normal, do not suffer from the disease themselves. Other cases may have no family history, meaning that the change in the X chromosome is a new one. This would be a mutation of the gene. Very rarely, a female with hemophilia is born if her mother is a carrier and her father has hemophilia. DNA testing can establish that a female is a carrier of the disorder. In the event of pregnancy, she may then have tests, such as CVS or amniocentesis, to determine the sex of the baby. A male fetus would have a 50-50 chance of of inheriting hemophilia.

Fragile X Syndrome This is an inherited abnormality of the X chromosome which causes intellectual problems ranging from mild learning disabilities to severe mental retardation. It is the leading known cause of genetically linked mental retardation. Most individuals with Fragile X have physical characteristics which can be linked to connective tissue disorder. The three most common physical features are large testicles, large ears, and a long narrow face. Approximately 80% of adult Fragile X males have one or more of these features. Severe speech and language delays are seen in Fragile X individuals, along with unique behavioral, and attentional characteristics. It is believed that these characteristics are the result of sensory integration dysfunction. The brain in affected individuals does not process or organize the flow of sensory impulses correctly. The Fragile X syndrome is called an X-linked disorder because its gene is located on an X chromosome. It is estimated that one in 1000 individuals are affected and about one in 700 females are carriers. Many cases of Fragile X are not the result of a new mutation and may be passed from generation to generation. Female are not affected as often or usually as severely if they carry a Fragile X since their normal X acts as a compensator. Conversely, since males have one X and one Y, they are almost always affected. Presently there is no known cure for Fragile X, however, individuals do benefit from various forms of special education. The test for Fragile X is different than the normal chromosome testing. Both the Fragile X test and the regular chromosome test need to be done as sometimes the individual does not have Fragile X but another chromosomal abnormality. A molecular DNA test is 790% accurate in detecting Fragile X.

INHERITED DISORDER TYPES

Dominant Genetic One affected parent has a single faulty gene (D) which dominates its normal counterpart (d). Each child produced has a 50% risk of inheriting the D, and the disorder from the affected parent.

X-Linked Genetic T he mother, who has a defective gene on one of her two sex chromosomes, is protected against the defect because her normal sex chromosome (x) compensates for the defect on the other (X). The father has normal male sex chromosomes (x and y). Each male child has a 50% risk of inheriting the faulty X, and the disorder, and a 50% chance of inheriting the normal x chromosome. Each female child has a 50% risk of inheriting the faulty X and becoming a carrier like her mother, and a 50% chance of inheriting two normal X chromosomes.

Recessive Genetic Both parents, usually unaffected, carry a defective gene (g) but are protected by the presence of a normal gene (G) which is generally sufficient for normal function. Each child produced has a 25% risk of inheriting a double dose of the g gene, which may cause a serious genetic defect; a 25% chance of inheriting two normal genes; and a 50% chance of being a carrier like both parents.

PRENATAL TESTING

Ultrasound This is a technique that uses sound waves to take a picture of the baby in the womb. It is used to locate the position, size and structure of the fetus and placenta in the womb. It is valuable in determining the age of a fetus and can detect multiple pregnancies as well as many malformations such as spina bifida, and various heart or kidney problems. Because it uses sound waves instead of radiation, ultrasound is safer than X-rays. It works by bouncing sound waves off the developing fetus. Echoes from the waves are converted into

an image on a TV monitor. Ultrasound may be used to help perform other prenatal diagnostic tests. If amniocentesis or CVS are called for, doctors are guided by ultrasound in removing cells to test for certain possible birth defects. For pregnancy scanning, sound waves are usually sent by a hand held device rubbed back and forth across the pregnant woman's abdomen. Depending on the position of the fetus and the purpose of the ultrasound, the procedure may take as long as an hour. Information obtained using ultrasound is often used to alter prenatal care to improve the chances to deliver a healthy baby. Ultrasound and other prenatal tests can let a woman know if her baby has certain birth defects or other special risks. Knowing about the problems before birth provides time to plan the baby's treatment if needed.

CVS Chorionic villus sampling is a prenatal test that can diagnose or rule out certain birth defects. The test is generally performed about the 10th or 11th week of pregnancy. CVS is not routinely offered to all pregnant women because the test carries a risk of miscarriage, and possibly other complications. CVS may be offered when there is an increased risk of chromosomal or genetic birth defects and parents would like test results as early in pregnancy as possible. CVS requires taking a small piece of the chorionic villi. Either a needle is inserted through the abdomen or a slim tube is inserted through the vagina to take a tiny tissue sample from outside the sac where the baby develops. The tissue is analyzed for chromosome disorders or various genetic conditions. Results are usually ready in one to two weeks. CVS tests are slightly less accurate than amniocentesis for chromosome analysis, and the risk of miscarriage is higher— one in 50 to 100.

Amniocentesis This is one of the best-known prenatal diagnostic techniques for certain congenital disorders. It is usually performed between the 13th and 15th weeks of pregnancy. In this procedure, a thin, hollow needle is inserted into a woman's uterus through the abdomen, guided by ultrasound used to view the fetus. The technician draws out some fluid surrounding the fetus. Fetal cells floating in the fluid can be analyzed to detect chromosomal abnormalities, more than 100 metabolic disorders and some anatomic defects. There is low risk (less than one in 200) of miscarriage or infection following the procedure. Results of the test are ready in one to three weeks.

Alpha-Fetoprotein Screening This blood test is most often done between 16 and 18 weeks. The results are usually available within a week. The test identifies pregnancies at higher-than-average risk of certain serious birth defects, such as spina bifida and Down syndrome. The test can provide information about a developing fetus. In most cases, an abnormal test result does not indicate a problem with the fetus. Alpha-fetoprotein(AFP) is a substance produced by the liver of the fetus. A small amount of AFP passes into the mother's bloodstream, where the concentration rises gradually. AFP levels can be measured during pregnancy by taking a sample of either the mother's blood or the amniotic fluid. If the test levels are abnormal, the test may be repeated. This test cannot diagnose a birth defect, it can only indicate an increased risk. Neural tube defects are among the most common and severe problems associated with high test levels. Low test level results are sometimes associated with chromosomal abnormalities, such as Down syndrome. However, for the majority of women, this test provides reassurance that their fetus does not appear to have certain serious birth defects.

Umbilical Vein Sampling (Cordocentesis) In this procedure a fine needle is passed through the mother's abdomen into the fetal vein in the umbilical cord. The technique allows fetal blood to be tested, facilitates intrauterine blood transfusions, and enables drugs to be injected directly into the baby if necessary

PKU All states screen newborns for phenylketonuria. This was the nation's first newborn screening test, and it has been routinely administered since the 1960s. PKU affects one baby in 12,000. Babies with this disorder cannot process a part of protein called phenylalanine. As a result, without treatment, phenylalanine builds up in the bloodstream and causes brain damage and mental retardation. When this disorder is detected early, mental retardation can be prevented by feeding the baby a special formula. A special diet should be followed, perhaps indefinitely.

Hypothyroidism The disorder most commonly identified by routine screening is congenital hypothyroidism, which affects one baby in 4,000. All states test newborns for this disorder. This is a thyroid hormone deficiency that retards growth and brain development. If it is detected in time, a baby can be treated with oral doses of thyroid hormone to permit normal development.

AIDS The AIDS screening test, which detects HIV antibodies, is not reliable for an infant born to an infected mother. This is because the mother's antibodies may be present in her baby's blood for up to 15 months, even if the baby has not been infected. A new test that measures an immune system protein called lgA is proving accurate in diagnosing HIV in babies between the ages of 3 and 6 months. Researchers also recently reported the development of a test that detected the virus in 80% of infected newborns. There is evidence that medicating a pregnant woman who tests positive for HIV can reduce the risk of her baby becoming infected. Much research in this area is being done at this time. AIDS screening for both mother and child remains voluntary. Infants with AIDS are closely monitored and treated with antibiotics, immune globulin, and anti viral drugs.

Galactosemia All states test newborns for galactosemia, which affects one baby in 60,000 to 80,000, and can cause blindness and mental retardation. A baby with galactosemia is unable to convert galactose, a sugar present in milk, into glucose, a sugar the body is able to use. As a result, milk and other dairy products must be eliminated from the baby's diet.

Sickle Cell Anemia In 1987, a National Institutes of Health panel recommended that all babies be screened for sickle cell anemia, an inherited blood disease. Currently, more than 30 states have begun screening for this disorder, which affects about one in 400 black babies in this country and also occurs with some frequency among people of Hispanic, Mediterranean, Middle Eastern and South Asian descent. Early treatment can prevent some of the complications of the disease. Young children with sickle cell anemia are especially prone to certain dangerous bacterial infections, such as pneumonia and meningitis. Studies have shown that treatment with penicillin, beginning by two months and continuing to about five years, can dramatically reduce the risk of these infections and the deaths that result from them. Newborn screening can alert the physician to begin antibiotic treatments before infections occur.

Cystic Fibrosis Several states now test for cystic fibrosis, a disease which occurs in up to one in 2,000 white births and produces chronic respiratory disease, problems with digestion, and poor growth. Infections involving the lungs are especially serious, and some may be prevented with antibiotics. Recent improvements in treatment have led to longer and healthier lives for children with cystic fibrosis. Experimental gene therapy has begun for individuals with cystic fibrosis and may someday present a cure for affected children.

Adrenal Hyperplasia A few states have begun testing for congenital adrenal hyperplasia. This disorder, in which certain hormones are deficient, affects genital development and can result in death in the newborn

period due to loss of salt from the tissues. Lifelong treatment with the missing hormone suppresses this disease, which occurs in one in 12,000 births.

Biotinadase Deficiency A fairly recent newborn screening test detects biotinadase deficiency. Biotinidase is an enzyme that recycles biotin, a B vitamin, in the body. A deficiency of this chemical, which occurs in one in 70,000 babies, may cause serious complications and even death. If the deficiency is detected in time, problems can be prevented by giving the baby extra biotin.

ADDITIONAL RISK FACTORS

Toxoplasmosis Toxoplasmosis is a parasitic infection that, when contracted by a pregnant woman, can pose a serious risk to her unborn baby. Up to one in 1,000 babies in this country are born infected with toxoplasmosis. A pregnant woman who contracts toxoplasmosis for the first time during pregnancy has about a 40% chance of passing the infection on to her fetus. Babies whose mothers had toxoplasmosis in the first trimester usually have the most severe infections. Up to 90% of infected babies appear normal at birth. Yet, 80 to 90% will develop sight-threatening eye infections months to years after birth. Some will also develop hearing loss, hydrocephalus, mental retardation, learning disabilities or seizures. Toxoplasmosis during pregnancy also can result in miscarriage or stillbirth. About one in 10 infected babies has a severe Toxoplasma infection that is evident at birth. These newborns have severe eye infections and other complications. Toxoplasmosis is caused by a parasite. It is most often picked up through exposure to cat feces or by eating raw or undercooked meat that is contaminated with the parasite. Other sources of infection may include raw goat's milk, raw eggs, and insects such as flies and cockroaches that may have been in contact with cat feces. Toxoplasmosis is one of the most common infections in the world. Most cases go undiagnosed. Symptoms tend to resemble the flu. About 60 to 85% of American women of childbearing age have never had toxoplasmosis and are susceptible to it during pregnancy. If it is suspected that a pregnant woman has an active case, a blood test that detects antibodies can be done. If the woman has an active infection, the fetus is tested also. Studies show that prenatal tests including amniocentesis and ultrasound can usually tell if the fetus is infected. Infected babies should be treated as soon as possible after birth with drugs which can help prevent or reduce the disabilities associated with toxoplasmosis.

Alcohol Each year approximately 5,000 babies , one in every 750, are born with fetal alcohol syndrome (FAS), a combination of physical and mental birth defects. In addition, almost 50,000 babies are born each year with fetal alcohol effects (FAE), a condition characterized by some, but not all, of the birth defects associated with FAS. Among the known causes of mental retardation, FAS is one of the most common and is the only cause that is entirely preventable. Babies with FAS are abnormally small at birth and usually do not catch up as they get older. Most of them have small eyes, a short upturned nose and small, flat cheeks. Their organs, especially the heart, may not form properly. Most babies

with FAS also have a small brain and some degree of mental retardation. Many have poor coordination, a short attention span, and exhibit behavioral problems. The effects of FAS last a lifetime. No level of drinking has been proven safe. Even moderate amounts of alcohol may damage the fetus. The fetal brain and other organs begin developing around the third week of pregnancy and are vulnerable to damage in those early weeks. Because no amount of alcohol is proven safe, women should stop drinking immediately if they suspect that they might be pregnant. This refers to all types of alcohol, including wine, wine coolers, beer and mixed drinks. Consuming alcohol during pregnancy increases the risk of miscarriage, stillbirth, and death in early

infancy. Because there currently is no way to predict which babies will be damaged by alcohol, the safest course is to not drink during pregnancy.

Smoking Smoking during pregnancy is associated with low birth weight, high rates of SIDS (sudden infant death syndrome), behavior problems, and learning difficulties. It is believed that smoking reduces the flow of oxygen and nutrients to the fetus. There is an increased risk of miscarriages and stillbirths in women who smoke while pregnant. Babies and children who are around smokers have more colds, ear infections, and flu. Children whose parents smoke are more likely to grow up to be smokers.

Cocaine T his is among the most dangerous drugs to unborn babies. It is estimated that more than 100,000 cocaine-exposed babies are born each year in the United States and that there may be as many as four million cocaine-exposed children by the year 2000. Cocaine use during early pregnancy can cause miscarriage. When the drug is used late in pregnancy, it may trigger labor. It can also cause a stroke or death to the unborn baby. Studies show that women who use cocaine during pregnancy are three times more likely to have a premature baby. Cocaine cuts the flow of nutrients and oxygen to the baby. Cocaine-exposed babies tend to have smaller heads, which may indicate a smaller brain. Cocaine use can also cause the placenta to pull away from the wall of the uterus before labor begins. This condition can be fatal for both mother and baby. Many exposed babies are born too soon or too small. Some studies suggest that cocaine-exposed babies are at increased risk of birth defects. It has been reported that mothers who used cocaine early in pregnancy were five times as likely to have a baby with a malformation of the urinary tract. Babies whose mothers used cocaine during pregnancy often score poorly on tests given at birth. Babies exposed to cocaine before birth also may have respiratory and neurological problems, including seizures,during the newborn period. Beginning at birth, these babies go through withdrawal from the drug. Many are very jittery and irritable. Bonding between mother and baby is often difficult. Babies born to mothers who use drugs during pregnancy are at increased risk for child abuse and neglect.

Rubella This is a mild, infectious disease caused by a virus. The problem with rubella is the damage it can cause an unborn baby whose mother has it during pregnancy. The first three months are the most dangerous, but the fetus can still be affected by rubella after that time. The earlier in pregnancy that the mother has rubella, the more serious the birth defects usually are. Some of these babies die before or shortly after birth. More common are the lifetime defects that a baby may be left with. Among these are hearing loss or deafness, eye defects, heart defects, mental retardation, and behavior problems. There is a blood test to find out if a person has rubella or has had it at some time. If the test shows rubella antibodies are present, then the person has had the disease and can’t get it again. This test is important for females of childbearing age. There is a vaccination against rubella which makes people immune to the disease. Rubella vaccine is recommended for all children, most teenagers, and many adults. Vaccinating girls as soon as they reach childbearing age protects their future babies. Women of childbearing age should be vaccinated only if they are sure they are not pregnant and don’t expect to be for the next three months.

STD Sexually transmitted diseases are infections which are caused by engaging in sex with someone who has one of the diseases. It is estimated that 12,000,000 people contract STDs each year. The deadliest STD is AIDS. Other common STDs are syphilis, gonorrhea, chlamydia, genital herpes, and genital warts. All STDs pose a risk to a pregnant woman and her unborn child. If the woman is infected with AIDS the fetus can contract the virus from the mother during pregnancy or delivery. A baby can catch chlamydia during a vaginal birth, causing ear and eye infections, and possible pneumonia. Genital herpes in newborns can cause severe skin infections, nervous system damage, blindness mental retardation or death. Gonorrhea in a baby can cause serious eye infections and blindness. Syphilis can be passed on to the fetus, causing damage to the heart,

blood vessels and nervous system, blindness, and death. Genital warts may be contracted by a baby during birth, causing wart growth inside the voicebox and blocking the windpipe.

Stress Stress can result from the physical and emotional changes of pregnancy. It causes changes in the body, including increased muscle tension, faster breathing, quicker heartbeat and increased blood pressure. How a pregnant woman deals with stress can make a difference in her physical and emotional well-being. It may even affect her unborn baby. Shifts in hormone levels also influence a woman's psychological state. There is evidence that high levels of stress can adversely affect pregnancy. Some studies suggest a possible relationship between stress and premature labor and low birthweight. Stress also appears to play a role in pregnancies complicated by high blood pressure or diabetes, conditions that also increase the risk of premature labor. Women with a combination of high stress levels and medical risk factors have a risk of premature labor. Not all studies have found a link between stress and low birthweight. Factors that affect an individual's ability to handle stress include personality, physical and mental health, living conditions, economic status, maturity and support. Stress management techniques such as meditation and other self-awareness programs can help alleviate stress. Walking and a regular exercise program are good options . One of the best ways to help prevent stress is to obtain early, comprehensive and continuous prenatal care.

Low Birthweight Low birthweight affects one in every 15 babies born each year in the United States. It is related to approximately 60% of infant deaths. These babies may face serious health problems, and are at increased risk of long term disabilities. Low birthweight is a weight of 5 pounds, 8 ounces or less at birth. Very low birthweight is a weight of 3 pounds, 5 ounces or less. Preterm births occur before the 38th week of pregnancy. Most low-birthweight babies are preterm. Small-for-date babies may be full-term but are underweight. The mother's medical problems influence birthweight, especially if she has high blood pressure, diabetes, certain infections or heart, kidney or lung problems. Smokers have smaller babies than nonsmokers. Drug and alcohol use limits fetal growth. Women under 17 years old or over 35 are at increased risk of having low- birthweight babies. A premature baby may have breathing problems. Up to 75% of babies born before the 30th week suffer from respiratory distress syndrome, a leading cause of death and disability among premature babies. Some low-birthweight babies have salt or water imbalances or low blood sugar which can cause brain damage. A premature baby may be anemic. Infants born too soon may not have had enough time to store iron. Low-birthweight babies may not have enough fat to maintain a healthy body temperature. Bleeding in the brain, which can be one of the most severe results of low birthweights, happens in 40 to 45% of very low-birthweight infants. The bleeding may result in brain damage or death. Premature babies often have a potentially dangerous heart problem. Lifesaving equipment in intensive care nurseries helps sustain low-birthweight babies who otherwise might not survive. The most important prevention is early and regular prenatal care. Women who receive this care can learn good health habits and ways to reduce the risk of having a low-birthweight baby.

Diabetes This is a disorder in which the body does not produce enough insulin or does not utilize insulin properly. Without treatment, high levels of sugar can accumulate in the blood and damage organs, including blood vessels, eyes, and kidneys. About one in 1,000 pregnant women has had diabetes before pregnancy. Another 3 to 12% develop diabetes for the first time during pregnancy. This is called gestational diabetes and is one of the most common complications of pregnancy. Babies of all diabetic women are at increased risk of health problems arising during the newborn period, including respiratory distress, low blood sugar and calcium levels, and jaundice. Early and regular prenatal care can reduce these risks. The American Diabetes Association recommends that all pregnant woman be screened for gestational diabetes. The screening test is usually done between the 24th and 28th week of pregnancy. All pregnant women with diabetes should follow a special diet and exercise, according to their doctor's recommendations.

Chicken Pox and Fifth Disease These diseases can sometimes pose a risk to the fetus if the mother contracts them during pregnancy. If she has been exposed to either of these illnesses, it is important that she inform her doctor. A blood test can usually determine if a person is susceptible to chicken pox. Although most women are immune because they have had the disease, one to five women in 10,000 will come down with chicken pox during pregnancy. This may cause a pattern of birth defects in the fetus in a small number of cases. Chicken pox also may increase the risk of miscarriage and premature labor. When maternal infection occurs around the time of birth, infection in the newborn can often be prevented or lessened if the baby is treated promptly with special medication. Fifth disease in pregnancy has not been proven to cause birth defects. It can, however, disrupt the fetus's ability to produce red blood cells. Sometimes this leads to a dangerous form of anemia, heart failure, abnormal pooling of fluid, and fetal death. Fewer than one-third of infected mothers pass this infection on to their babies. There is no vaccine to prevent fifth disease or to cure it. A pregnant woman who has been exposed to fifth disease should consult her doctor promptly.

Teaching The Unit

Goal Students will demonstrate academic progress and developmental growth in scientific knowledge.

Objectives Students will expand their knowledge in the specific areas of:
1. Genetics

2. Birth disorders

3. Pregnancy

Methodology Students will gain an understanding of the unit topics in a variety of ways, according to their individual learning styles and ability levels. Materials will be presented in the following manner:

1. Individual student reading of written, factual information relating to the unit
2. Group reading of information on topics of specific interest to the group
3. Teacher lecture and demonstration to inform and clarify unit topics
4. Whole class, and small group discussions of particular portions of the unit
5. Experiential, hands-on projects, such as building a cell model, involving all class members, either individually or in small groups

Evaluation Knowledge and understanding, of the topic units, gained by the students will be evaluated by using the following criteria:

1. Teacher corrected worksheets
2. Graded tests

3. Completed assignments
4. Teacher observation of group project participation, interaction and results
5. Teacher observation of individual involvement and contribution to group discussions
6. Teacher observation of student interest and effort maintained in completing individual projects

Sample Activity Unit Topic, Genetics

This worksheet to be used following study of the unit topic: *Background/Mendel.*

Directions : Choose the best answer from the box. Write that answer in the blank.

recessive	pea plants
Gregor	Mendeltall
two	female
male	genes

1. _____ is often called the father of genetics.
2. He worked in a garden and experimented with _____ .
3. He observed that the _____ gene is dominant in pea plants and the short gene is _____ .
4. Today Mendel's factors are called _____ .
5. One gene comes from the _____ reproductive cell and the second one comes from the _____ cell.
6. Organisms receive _____ genes for each trait.

Sample Project Unit Topic, Genetics

To be completed following study of the unit topics: *Cells, Chromosomes, Genes, and DNA.*

Directions : Students will construct individual cell models in the following manner:

1. Air filled balloons, of various shapes and sizes, will be used as base forms.
2. These forms will be covered with papiermache' - torn strips of newspaper dipped in wheat paste.

3. When dry and hard, these shapes may be painted.
4. Using an xacto knife, they may be split in half horizontally.
5. These models may then be filled with materials representing cell matter (the nucleus, protoplasm, chromosomes, etc.).
6. The cell forms may be “hinged” on the inside using duct tape , enabling them to be easily opened and closed.
7. The completed cell models might be displayed in the classroom or in an appropriate location within the school building, such as the library or a hallway display case.

Sample Project Unit Topic, Pregnancy

This activity should be completed following study of the unit topic: *Prenatal Testing* .

Directions : Students, working together, will create a pregnancy timeline in the following manner:

1. Tape or staple together nine pieces of colored paper (ex. pink, blue, yellow) alternating the three colors, to represent the three trimesters of pregnancy.
2. Indicate week 1, 2, 3 etc. on the timeline by taping numbers across the top of the strip. (1-40)
3. Using index cards, label and write a brief description of the following prenatal tests:
   Amniocentesis 13-15 weeks
   Chorionic Villi Sampling (CVS) 9-12 weeks
   Alpha-Feto Protein Screening (AFP) 15-18 weeks
   Ultrasound may be used throughout, more often during midterm
4. Tape the prenatal test cards to the timeline in the appropriate section.
5. Display this “work-in-progress” in the classroom.
6. Individual students may add their own prenatal testing dates, results or comments, as they feel comfortable.

Sample Test Unit Topic, Birth Disorders

This test should be completed following study of the unit topic: *Birth Disorders.*

Directions : Match the disorder with the sentence. Write the correct answer on the line.

1. Babies with this disorder cannot process a part of protein called phenylalanine _____ .
2. This is a thyroid hormone deficiency that retards growth and brain development _____ .
3. The major cause of this disorder is an extra chromosome 21 _____ .
4. This is a birth defect of the backbone and sometimes the spinal cord _____.
5. This disease occurs most frequently in descendants of Central and Eastern European Jews _____ .
6. People affected with this disorder are often tall, slender, and loose-jointed _____ .
7. This disease affects a protein inside the red blood cells _____ .
8. It is a genetic disorder of bone growth that is evident at birth _____ .
9. This muscle-wasting disorder affects boys almost exclusively _____ .
10. The body produces large amounts of abnormally thick mucus because of this disease _____ .

muscular dystrophy PKU sickle cell disease

Down syndrome hypothyroidism Tay-Sachs spina bifida

achondroplasia Marfan syndrome cystic fibrosis

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