IV. LEARNING ACTIVITIES
C. EXERCISE I:

TASK: Student will read the text and follow the information in the prospectus and guidelines in the operational rules to demonstrate his/her knowledge of the skill and information.

TEXT: Problems in Prenatal Development
In the United States, about 97% of mothers deliver infants that are healthy and problem-free, a phenomenon that illustrates just how remarkably and predictable the sequence of prenatal development is. However, this sequence of development is not immune to modification or outside influences, as you will soon see in detail.

The potential problems fall into two general classes: genetic and chromosomal problems that begin at conception, and problems caused by damaging substances or events called teratogens.

Genetic disorders

Genetic traits can be passed through families in several distinct patterns. The most common patterns are the following: Dominant genetic diseases are caused by a mutation in one copy of a gene. If a parent has a dominant genetic disease, then each of that person’s children has a 50% chance of inheriting the disease.

What Is Autosomal Recessive Disease?

Some health problems are passed down through families. There are different ways this can happen. To have a child born with what’s called an “autosomal recessive disease” like sickle cell disease or cystic fibrosis, both you and your partner must have a mutated (changed) gene that you pass on to your child.
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   How Are People Affected?

Almost every cell in your body contains 23 pairs of tightly wound DNA called chromosomes. You get 23 of them from your mother and 23 from your father.

One pair of chromosomes decides your sex. The others contain thousands of different genes that decide every other trait you have, from hair and eye color to your risk of getting diseases. These are called autosomes.

Some genes are "dominant." You only need one from a parent to have that trait. Other genes are "recessive."

With them, you must inherit the same gene from both parents to be affected.

If one of your parents passes on a recessive gene to you that can cause disease, then you become a "carrier." You likely won't have any symptoms since the other gene is normal. In fact, many people won't know they're a carrier without being tested.

But if you and your partner both have the same mutated gene, there's a 25% chance that your child will be born with a severe disease.

Anyone can carry a recessive gene that causes illness, but some diseases are more common in certain ethnic groups.

What Are Some Types of These Diseases?

Common autosomal recessive disorders include:
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- **Sickle cell disease**: About 1 in 12 African American people are carriers of this disease. One in 500 African American babies is born with it. Sickle cell causes your red blood cells to be stiff and sticky so they can’t easily move oxygen through your body. This puts you at risk for painful complications and severe infections.

- **Cystic fibrosis (CF)**: People with this disorder produce very thick mucus that sticks to their lungs and harms major organs. CF also makes it hard for your body to digest and absorb food.

- **Tay-Sachs disease**: This causes intense damage to the central nervous system. It occurs most often in people whose ancestors are Ashkenazi Jews, French Canadian, Amish, or Cajun.

- **Gaucher disease**: Many of your organs and tissues can be damaged from this disease. An enlarged liver and spleen, as well as anemia, are common. Some people also have seizures and brain damage. The most severe type causes problems for babies before they’re born or in the days right after birth.

Many autosomal recessive diseases will have a severe effect on the life of a child. In some cases, they may be fatal.

**AUTOSOMAL DOMINANT INHERITANCE**

One of the ways a genetic trait or a genetic condition can be passed down (inherited) from parent to child. In autosomal dominant inheritance, a genetic condition can occur when the child inherits one copy of a mutated (changed) gene from one parent. A child who has a parent with the mutated gene has a 50% chance of inheriting that mutated gene.
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Autosomal dominant inheritance is a way a genetic trait or condition can be passed down from parent to child. One copy of a mutated (changed) gene from one parent can cause the genetic condition. A child who has a parent with the mutated gene has a 50% chance of inheriting that mutated gene. Men and women are equally likely to have these mutations and sons and daughters are equally likely to inherit them. Disorders caused by dominant genes, such as Huntington’s disease Are not usually diagnosed until adulthood. This disorder causes the brain to deteriorate and affects both psychological and motor functions.

Sex Linked Disorders

Most sex-linked disorders are cause by recessive genes. Sex linked is a trait in which a gene is located on a sex chromosome. In humans, the term generally refers to traits that are influenced by genes on the X chromosome. This is because the X chromosome is large and contains many more genes than the smaller Y chromosome. In a sex-linked disease, it is usually males who are affected because they have a single copy of X chromosome that carries the mutation. In females, the effect of the mutation may be masked by the second healthy copy of the X chromosome.

These are traits that are found on either one of the chromosomes that determine sex, or the sex chromosomes. And in humans this is the X or the Y chromosomes. Some of the more familiar sex-linked traits are hemophilia, red-green color blindness, congenital night blindness, some high blood pressure genes, Duchenne muscular dystrophy, and also Fragile X syndrome. What’s also very interesting is that you can imagine that for individuals who are XY or males, having these different mutations on the genes, on the X chromosome, is particularly problematic, because unlike females, there are not two X chromosomes that
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give you the potential of carrying a normal gene on the X chromosome. Which is why in many cases you'll see that males are more often afflicted with these sex-linked disorders.

Trisomy’s

Genes are the blueprint for our bodies. Almost every cell in the body has a copy of the blueprint, stored inside a sac called the nucleus. Genes are beaded along chromosomes, which are tightly bundled strands of the chemical substance deoxyribonucleic acid (DNA). Humans usually have 23 pairs of chromosomes, with two sex chromosomes that determine sex and 44 chromosomes that direct other factors, such as growth and function.

A chromosome condition is caused by an alteration in the number or genetic structure of chromosomes.

Trisomy (‘three bodies’) means the affected person has three copies of one of the chromosomes instead of two. This means they have 47 chromosomes instead of 46.

Down syndrome, Edward syndrome and Patau syndrome are the most common forms of trisomy. Children affected by trisomy usually have a range of birth anomalies, including delayed development and intellectual disabilities.
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CHROMOSOME ANOMALIES

Most humans have 46 chromosomes in their cells, which occur in pairs for a total of 23. Twenty-two of these pairs are quite similar in both males and females, but the final pair is the sex chromosomes. This occurs typically as “XX” in women and “XY” in men. When there are differences in these chromosomes from the usual presentation, these are known as sex chromosome anomalies. Sex chromosome anomalies are caused by genetic mutations. These anomalies can either be passed along from parents to children or occur at random for unknown reasons.

Symptoms can vary widely depending on what type of sex chromosome anomaly is present. They can range from having the sexual and physical characteristics of the opposite gender to having more prominent male or female characteristics to having mental or physical disabilities, among many other symptoms.

There is no cure for sex chromosome anomalies. In some cases, hormone therapy, reconstructive surgeries or other forms of therapy can help resolve some of the issues related to sex chromosome anomalies.