

Genetic and Developmental Diseases

OBJECTIVES/RATIONALE

The effects of genetic diseases have life-long consequences. Although some genetic and developmental disorders may first emerge at birth, these disorders may appear at any age. The student will identify common genetic and developmental disorders, their important signs and symptoms and common tests used to diagnose these disorders.

TEKS: 121.15.(c) 3(D), 4(A-E), 5(A-D), 6(A-D), 7(C)

TAKS ELA 1, 4
Science 1,2,3

KEY POINTS

POWER POINT

I. Mitosis and Meiosis

- A. All cells of normal mature individual have 46 chromosomes.
 - a. These cells duplicate themselves and divide to form daughter cells, each with 46 chromosomes
 - b. Process is called **mitosis** and can occur with most cells in the body
- B. Germ cells that develop into sperm and ova undergo a different type of cell division called **meiosis**.
 - a. One chromosome from each pair is passed on to each gamete (sperm or ovum).
 - b. Each gamete has only 23 chromosomes.
 - c. When an ovum is fertilized with a sperm, the newly formed individual will have a combined total of the normal forty-six chromosomes (one half (23), from each parent).

II. Autosomal and Sex Chromosomes

- a. 44 of the 46 chromosomes determine body function - these are referred to as **autosomes**
 1. XX chromosomes = female
 2. XY chromosomes = male
 3. It is the male sperm that determines sex of fetus
- c. the sex chromosomes are in every cell of body and are responsible for directing activity of cell specifically for a female or for a male

III. Visualizing Chromosomes

- a. **Karyotyping** process to visualize chromosomes which involves:
 - i. taking picture of cell during mitosis
 - ii. arranging chromosome pairs in order from largest to smallest
 - iii. numbering chromosome pairs one through 23
- b. Sex chromosomes can be evaluated by a **buccal smear** -

Barr bodies

DNA

- each unit of DNA is called a
- ii. each chromosome is made up of thousands of genes located at precise positions in chromosome
- iii. chromosomes (one from each parent) pair up during fertilization of egg (alleles)
- iv. this matched gene pair determines heredity (characteristics inherited from parents)
- v. besides facial features, hair and eye color, heredity is thought to play a part in many other processes:
 - a. development of plaque in arteries
 - b. obesity
 - c. alcoholism
 - d. some mental illnesses

IV. Understanding basic heredity

A. **Genotypes** are the genetic pattern of an individual.

- a. each gene in an allele (matched pair) of genes may be dominant or recessive.
 - i. Dominant genotypes expressed with capital letter (example: brown eyes = B)
 - ii. Recessive genotypes are expressed with smaller (example: blue eyes = b)
 - iii. if alleles in a pair match (BB or bb), they are said to be **homozygous**

**heterozygous
phenotype**

Homozygous pairs (dominant or recessive) will always express that trait (BB = brown eyes, bb = blue eyes, etc.)

- d. Heterozygous alleles will express the phenotype (trait) of dominant gene only. (Bb = brown eyes)
- e. Heterozygous pairs are said to be carriers of recessive disorders - recessive traits will not be expressed unless paired with another recessive gene

V. Abnormalities

A. may be due to chromosomal, genetic, or environmental factors, or combination of these

- a. major chromosomal abnormalities usually lead to spontaneous abortion of fetus
number placement

- Line of inheritance is easily followed from one generation to another
 - iii. Dominant genes will always be expressed whether homozygous or heterozygous
 - iv. Example of autosomal dominant disorder: polydactyly (excessive number of finger or toes)
2. Autosomal recessive

- i. Only seen when two recessive genes are paired
- ii. Each parent may be phenotypically normal or without sign of disorder but is a heterozygous carrier of the disorder
 - a. When each parent is heterozygous, chance of offspring having disorder is one in four
 - b. If one parent has the disorder, chances increase to one in two
 - c. If one parent is homozygous dominant, none of offspring will be affected
 - d. These disorders may skip generations before it is paired with another recessive gene and is expressed
 - e. Example of autosomal recessive disorder: cystic fibrosis
- 3. sex-linked dominant - these are more rare than recessive disorders and are easily recognized
- 4. sex-linked recessive
 - i. these disorders are typically carried by females and passed to males
 - ii. reason for this: recessive gene disorders on the X chromosome of female are overridden by dominance of normal gene on other X chromosome
 - iii. in males, the X disorder is expressed because there is no corresponding gene on the Y chromosome.
 - iv. X-linked disorders usually appear every other generation since they are passed mother to son (mother to son; son to daughters (who become carriers) - affected male is unable to pass this disorder to sons because male gives a Y chromosome to sons, not an X.
 - v. example of sex-linked recessive disorder: hemophilia

VIII. anomalies

- A. Approximately two percent of all newborns have congenital anomalies (birth defects).
 - a. 65% of congenital anomalies are (unknown cause)
 - b. 20% are genetic
 - c. 5% are chromosomal
 - d. 10% are environmental (maternal radiation, infection, drugs, alcohol, medications)

IX. Diagnostic Tests

- A. physical for affected individual
- B. of fetus (determines malformations of head, internal organs, extremities)
- D. (amniotic fluid analysis to determine genetic and chromosomal disorders)
- E. maternal blood analysis to observe abnormal fetal substances

X. Common musculoskeletal genetic/developmental disorders

: Muscular Dystrophy (MD)

: group of genetically inherited diseases characterized by degeneration of muscles; most common type is Duchenne's MD.

Etiology: genetic

Manifestations

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Diagnosis:

Prognosis:

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Name
Description

Etiology:

Manifestations

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Diagnosis:

Treatment:

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ACTIVITIES

- I. Complete a case study on one of the following: Club Foot (Talipes Equinovarus), Osteogenesis Imperfecta, Spina Bifida, Patent Ductus Arteriosus, Coarctation of Aorta, Tetralogy of Fallot, Sickle Cell Anemia, Hemophilia, Hirschsprung's Disease, Phenylketonuria (PKU), Wilms' Tumor, Cryptorchidism, Turners Syndrome, Klinefelter's Syndrome, Cystic Fibrosis, Down Syndrome, Fetal Alcohol Syndrome, Congenital Rubella Syndrome, Anencephaly, Achondroplasia, Tay-Sachs Disease

MATERIALS NEEDED

Key Terms

Key Terms Answers

Quiz Key

<http://www.kumc.edu/gec/support/> - site for *Genetic and Rare Conditions*

For reinforcement, the student will define the Key Terms.

For enrichment, the student will research and report over one of the online genetic diseases (see Materials Needed). Present using multimedia technology.

REFLECTIONS

Pathology Report

Disease
Alternate names
Definition
Etiology
Signs & Symptoms
Diagnostic Tests
Treatment
Complications
Prognosis

Key Terms:

Zygote –

Answers: Key Terms

Quiz Answers: Genetic/Developmental Disorders

a
b
b
a
a
b
b
a
a
a
b