

Genetic and Developmental Diseases

Course

Pathophysiology

Unit VII

Pathology

Across the Life

Span

Essential Question

What effect do genes have on the cause of disease?

TEKS

130.208 (c)

4A,C

6A,B,E

7B

Prior Student Learning

Pathophysiology

Basics

Anatomy and

Physiology

Immune System

Estimated time

3 hours

Rationale

The effects of genetic diseases have lifelong consequences. Although some genetic and developmental disorders may first emerge at birth, these disorders may appear at any age.

Objectives

Upon completion of this lesson, the student will be able to

- Analyze the relationship between genes and chromosomes
- Identify common genetic and developmental disorders, their important signs and symptoms, and the common tests used to diagnose these disorders
- Differentiate among congenital, genetic, and hereditary disorders, and give examples of each
- Define karyotype and explain how karyotypes are used in genetic counseling

Engage

More than 30,000 people in the United States have cystic fibrosis and more than 10 million people carry the Cystic Fibrosis trait. What does this mean? How would you find out if you are a carrier or not?

Key Points

I. Mitosis and Meiosis

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

II. Autosomal and Sex Chromosomes

- A. 44 of the 46 chromosomes determine bodily functions – these are referred to as autosomes
- B. The remaining 2 chromosomes determine the individual's sex
 1. XX chromosomes = female
 2. XY chromosomes = male
 3. It is the male sperm that determines the sex of the fetus
- C. The sex chromosomes are in every cell of the body and are responsible for directing the activity of the cells specifically for a female or for a male

III. Visualizing Chromosomes

- A. **Karyotyping** – a process to visualize chromosomes which involves
 1. Taking pictures of the cell during mitosis
 2. Arranging chromosome pairs in order from largest to smallest
 3. Numbering chromosome pairs 1 through 23
- B. Sex chromosomes can be evaluated by a **buccal smear** – this test is performed by obtaining epithelial cells from the buccal cavity of the mouth, staining the cell, and microscopically observing for X chromosomes (referred to as **Barr bodies**)
- C. Barr bodies – present when two X chromosomes are present (female)
 1. X chromosomes are much larger than Y chromosomes and carry more genetic information.
 2. The X chromosomes carry genes for female characteristics and other genes essential to life (blood formation, metabolism activities, and immunization)
 3. The Y chromosome is smaller and only carries genes related to masculinity
- D. Chromosomes – made of units of **DNA** (arranged in a specific order)
 1. Each unit of DNA is called a gene
 2. Each chromosome is made up of thousands of genes located at precise positions
 3. Chromosomes (one from each parent) pair up during fertilization of the egg
 4. This matched gene pair determines heredity (characteristics inherited from parents)
 5. Besides facial features (hair and eye color), heredity is thought to play a part in many other processes:
 - a. Development of plaque in the arteries
 - b. Obesity
 - c. Alcoholism
 - d. Some mental illnesses

IV. Understanding basic heredity

- A. **Genotypes** are the genetic pattern of an individual
 1. Each gene in an allele (matched pair) of genes may be dominate or recessive.
 - a. Dominate genotypes expressed with capital letters (example: brown eyes = B)
 - b. Recessive genotypes are expressed with lower-case letters (example: blue eyes = b)
 - c. If the alleles in a pair match (BB or bb), they are said to be **homozygous**
 - d. If the alleles do not match (Bb) they are said to be **heterozygous**
 2. Expression of a trait (blue eyes, brown hair, etc.) is called a **phenotype**
 3. Homozygous pairs (dominant or recessive) will always express that trait (BB = brown eyes, bb = blue eyes, etc.)

4. Heterozygous alleles will express the phenotype (trait) of the dominate gene only (Bb = brown eyes)
5. 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 combinations of these
 1. Major chromosomal abnormalities usually lead to spontaneous abortion of the fetus
 2. Chromosomal disorders are usually related to the number or placement of the chromosomes
 3. Chromosomes may fail to separate properly during cell division causing one daughter cell to have an extra chromosome while the other daughter cell has no chromosomes
 4. Abnormal number or structure of autosomal chromosomes is usually incompatible with life because these chromosomes carry a large number of essential genes
- B. The most common autosomal chromosomal disorder is Down Syndrome

VI. Two ways that people acquire an abnormal gene:

- A. Mutation of the gene during meiosis
- B. Passing of an abnormal gene from the parents (heredity) – genetic disorders are passed to offspring in four different ways: autosomal dominant, autosomal recessive, sex-linked dominant, and sex-linked recessive
 1. Autosomal dominant
 - a. Easily recognized because the presence of disorders identifies individuals with the dominant gene
 - b. The line of inheritance is easily followed from one generation to another
 - c. The dominant genes will always be expressed whether homozygous or heterozygous
 - d. An example of an autosomal dominant disorder is polydactyl (excessive number of finger or toes)
 2. Autosomal recessive
 - a. Only seen when two recessive genes are paired
 - b. Each parent may be phenotypically normal or without sign of disorder, but is a heterozygous carrier of the disorder
 - i. When each parent is heterozygous, the chance of offspring having the disorder is one in four
 - ii. If one parent has the disorder, chances increase to one in two
 - iii. If one parent is homozygous dominant, none of the offspring will be affected
 - iv. These disorders may skip generations before being paired with another recessive gene and expressed

- v. An example of an autosomal recessive disorder is Cystic Fibrosis
- 3. Sex-linked dominant – these are more rare than recessive disorders and are easily recognized
- 4. Sex-linked recessive
 - a. These disorders are typically carried by females and passed to males
 - b. The reason for this is recessive gene disorders on the X chromosome of the female are overridden by dominance of the normal gene on the other X chromosome
 - c. In males, the X disorder is expressed because there is no corresponding gene on the Y chromosome
 - d. X-linked disorders usually appear every other generation since they are passed from mother to son (mother to son; son to daughters, who become carriers) – the affected male is unable to pass this disorder to his sons because the male gives a Y chromosome to his sons, not an X
 - e. An example of a sex-linked recessive disorder is hemophilia

VII. **Congenital** anomalies

- A. Approximately two percent of all newborns have congenital anomalies (birth defects)
 - 1. 65% of congenital anomalies are **idiopathic** (cause unknown)
 - 2. 20% are genetic
 - 3. 5% are chromosomal
 - 4. 10% are environmental (maternal radiation, infection, drugs, alcohol, medications, etc.)

VIII. Diagnostic Tests

- A. Physical for the affected individual
- B. **Ultrasonography** of the fetus determines malformations of the head, internal organs, and extremities
- C. **Amniocentesis** – amniotic fluid analysis to determine genetic and chromosomal disorders
- D. Maternal blood analysis to observe abnormal fetal substances

IX. Common musculoskeletal genetic/developmental disorders

- A. **Name** – Muscular Dystrophy (MD)
 - 1. **Description** – a group of genetically inherited diseases characterized by degeneration of the muscles; the most common type is Duchenne's MD
 - 2. **Etiology** – genetic
 - 3. **Manifestations**
 - a. Onset is usually between the ages of two and five years
 - b. The pelvic and leg muscles are usually affected first
 - c. Leads to a characteristic waddling gait
 - d. Toe walking
 - e. Lordosis
 - f. Gower's maneuver (an unusual way of getting up from a squatting position due to weakened pelvic muscles)

- g. Bulking of muscle mass (esp. Gastrocnemius) due to fat and connective tissue deposits

4. **Diagnosis**

- a. Physical exam, muscle biopsy, electromyography

5. **Prognosis**

- a. There is no cure for MD – physical therapy and leg braces are effective in maintaining mobility and quality of life
- b. Affected children are usually confined to a wheelchair by age nine
- c. Life expectancy is usually in the late teens or early twenties
- d. Death is commonly due to respiratory or cardiac complications

B. **Name** – Congenital Hip Dislocation

- 1. **Description** – an abnormality of the hip joint resulting in the femoral head slipping out of the acetabulum; more common in girls
- 2. **Etiology** – maternal hormones relax the mother's pelvic ligaments during labor, thus relaxing an infant's joint ligaments
- 3. **Manifestations**
 - a. Asymmetrical folds of the affected thigh
 - b. Difference in leg length
 - c. Limited abduction of the affected leg
- 4. **Diagnosis** – a physical examination and hip joint X-ray
- 5. **Treatment**
 - a. Closed reduction (placing the femoral head in the acetabulum) and maintaining normal position by use of a cast for approximately two to three months
 - b. Surgical treatment may be required in older children

Activity

I. Complete a Pathology Report 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
- Turner's Syndrome
- Klinefelter's Syndrome
- Cystic Fibrosis
- Down Syndrome
- Fetal Alcohol Syndrome

- Congenital Rubella Syndrome
- Anencephaly
- Achondroplasia
- Tay-Sachs Disease

Assessment

Successful completion of the Genetic and Developmental Quiz
Pathology Report

Materials

Key Terms

Key Terms Answers

Quiz Key

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

Accommodations for Learning Differences

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.

National and State Education Standards

National Health Science Cluster Standards

HLC01.01 Academic Foundations Health care workers will know the academic subject matter required for proficiency within their area. They will use this knowledge as needed in their role. Describe the basic structures and functions of cells, tissues, organs and systems as they relate to homeostasis. Analyze the interdependence of the body systems as they relate to wellness, disease, disorders, therapies and care rehabilitation. Compare selected diseases/disorders including respective classification, causes, diagnoses, therapies, and care/rehabilitation to include biotechnological applications. Analyze body system changes in light of diseases, disorders and wellness.

TEKS

130.208 (c)(4)(A) Identify biological and chemical processes at the cellular level,

130.208 (c)(4)(C) Identify factors that contribute to disease such as age, gender, environment, lifestyle and heredity

130.208 (c)(6)(A) Describe on the nature of diseases according to etiology, signs and symptoms, diagnosis, prognosis and treatment options,

130.208 (c)(6)(B) Explore advanced technologies for the diagnosis and treatment of diseases,

130.208 (c)(6)(E) Differentiate between congenital disorders and childhood diseases

130.208 (c)(7)(B) Analyze the effects of stress and aging on the body.

Texas College and Career Readiness Standards

Science Standards

Nature of Science: Scientific Ways of Learning and Thinking

D. Current scientific technology

1. Demonstrate literacy in computer use
2. Use computer models, applications and simulations

E. Effective communication of scientific information

1. Use several modes of expression to describe or characterize natural patterns and phenomena. These modes of expression include narrative, numerical, graphical, pictorial, symbolic and kinesthetic
2. Use essential vocabulary of the discipline being studied

III Foundation Skills: Scientific Applications of Communication

A. Scientific Writing

1. Use correct applications of writing practices in scientific communication

C. Presentation of scientific/technical information

1. Prepare and present scientific/technical information in appropriate formats for various audiences

D. Research Skills/information literacy

1. Use search engines, databases and other digital electronic tools effectively to locate information
2. Evaluate quality, accuracy, completeness, reliability and currency of information from any source

Disease Report Template

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

Disease Report Rubric

Criteria	Possible Points	Points Awarded
Disease Correctly names the disease.	3	
Alternate Names If applicable, includes any alternate names for the disease or disorder.	2	
Definition Includes an in-depth discussion of the history and general description of the disease with interesting facts	10	
Etiology Includes the cause or origin of the disease or disorder.	15	
Signs & Symptoms Accurately describes common physical and medical symptoms.	15	
Diagnostic Tests Identifies tests performed to aid in the diagnosis or detection of the disease or disorder.	15	
Treatment Identifies the mode or course pursued for remedial ends for the disease or disorder.	15	
Complications Identifies any diseases or injuries that may develop during the treatment of the disease or disorder.	10	
Prognosis Includes the prediction of the probable course, outcome, frequency, and life expectancy of the disease or disorder.	10	
Bibliography Follows proper format and includes more than 3 sources.	5	
TOTAL	100	

Key Terms

Alleles –

Amniocentesis –

Autosomes –

Bar bodies –

Buccal smear –

Congenital anomaly-

DNA –

Gamete –

Gene –

Genotype –

Germ cell –

Heterozygous –

Homozygous –

Idiopathic –

Karyotyping –

Meiosis –

Mitosis –

Ovum –

Phenotype –

Sperm cell –

Ultrasonography –

Zygote –

Answers: Key Terms

Alleles – matched gene pairs which determine heredity

Amniocentesis – a test performed by aspirating amniotic fluid from the uterus to determine the health, sex, or genetic constitution of a fetus

Autosomes – the forty-four of the forty-six chromosomes, which determine body function

Barr bodies – inactive X-chromosome found in the nuclei of somatic cells of females

Buccal smear – a test where squamous epithelial cells are brushed from the buccal cavity of the mouth to evaluate sex chromosomes

Congenital anomaly – a birth defect

Dioxyribonucleic acid (DNA) – a nucleic acid molecule in the form of a twisted double strand (double helix) that is a major component of the chromosomes and carries genetic information

Gamete – germ cell; a specialized male (sperm) or female (ovum) cell with half the normal number of chromosomes that unites with another cell of the opposite sex during the process of sexual reproduction

Gene – the basic unit capable of transmitting characteristics from one generation to the next (consists of a specific sequence of DNA or RNA that occupies a fixed position on a chromosome)

Genotype – the genetic makeup of an organism, as opposed to its physical characteristics (phenotype)

Heterozygous – having different alleles for any one gene

Homozygous – having two identical genes

Idiopathic – a disease or disorder that has no known cause

Karyotyping – a process that involves taking a picture of a cell during mitosis, arranging the chromosome pairs in order from largest to smallest, and numbering them one through twenty-three

Meiosis – the process of cell division in organisms that reproduce sexually; the nucleus divides into four nuclei, each of which contains half the usual number of chromosomes

Mitosis – the process by which a cell divides into two daughter cells, each of which has same number of chromosomes as the original cell

Ovum – a female reproductive cell

Phenotype – expression of bodily traits (brown hair, blue eyes, etc.)

Sperm cell – spermatozoon; a male reproductive cell (gamete) that has an oval head with a nucleus, and a tail by which it moves to find and fertilize an ovum

Ultrasonography – the process of making a picture of internal organs with sound waves for diagnostic purposes

Germ cell – the smallest element in an organism that is capable of growing into a complete adult or part

Zygote – an ovum that has been fertilized by a spermatozoon

True/False Quiz: Genetic and Developmental Disorders

- _____ 1. Somatic cells have the ability to reproduce in a process called mitosis.
a. true b. false
- _____ 2. The normal number of chromosomes pairs is 46.
a. true b. false
- _____ 3. The onset of muscular dystrophy is between 12 and 18 years of age
a. true b. false
- _____ 4. Heterozygous pairs are said to be carriers of recessive disorders.
a. true b. false
- _____ 5. An expression of bodily traits is referred to as phenotype.
a. true b. false
- _____ 6. Congenital anomalies are always due to chromosomal defects.
a. true b. false
- _____ 7. Bar bodies are evidence of Y chromosomes.
a. true b. false
- _____ 8. *Germ* cells refer to sex cells: ova and sperm.
a. true b. false
- _____ 9. Genes on the chromosomes that align are called alleles.
a. true b. false
- _____ 10. Polydactyl is a sex-linked disease.
a. true b. false

Quiz Answers: Genetic/Developmental Disorders

1. **a**
2. **b**
3. **b**
4. **a**
5. **a**
6. **b**
7. **b**
8. **a**
9. **a**
10. **b**