

Sex-Linked Chromosomal Disorders

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Primary Learning Outcomes

Students will learn the names, characteristics, and causes of several different sex linked disorders, including muscular dystrophy. Students will create, describe and predict genotypes according to genetic pedigrees.

Additional Learning Outcomes

Students will be introduced to the ideas of genetic engineering and genetic counseling.

Materials

1. Paper
2. Colored pencils
3. Display board
4. Printer
5. Glue

Technology Connection

Students will need access to the Internet to complete research on a sex-linked chromosomal disorder.

Total Duration

2 hours, 50 minutes

Procedures

Step 1

Duration: Varies

Teacher Preparation

Review the lecture notes in Step 2 to become familiar with the concepts and assure that it is at the right level for the students. Students need prior knowledge of genetics terminology like dominant, recessive, carrier, sex-linked, as well as familiarity with pedigree symbols. A complete list is included in the "Genetics Terminology Word List". If they are not familiar with genetics terms, please use the "Definitions for Sex-linked Disorder Vocabulary" to help acquaint them. Additionally, a key to pedigree symbols is included in the "Pedigree Symbols" document for the students to review if needed. Both the vocabulary sheet and the pedigree symbols document can be passed out before the lecture. Refer to the Web resources below for additional information about genetics terminology and pedigree symbols.

Web Resources

Title: Basic Principles of Genetics

URL: <http://anthro.palomar.edu/mendel/Default.htm>

Description: This is a tutorial Web site on the basics of genetics. This site includes vocabulary with definitions as well as links to other related information.

Title: Pedigree symbols and Genetic Terms

URL: <http://www.people.virginia.edu/~rjh9u/pedsymb.html>

Description: This Web site contains some additional pedigree symbols and genetic terms with definitions. It includes symbols for twins and still births.

Title: Pedigree symbols, rules, guidelines and examples

URL: <http://www.rhjh.lkwash.wednet.edu/homehelp/familytree/>

Description: This Web site contains pedigree symbols, rules, guidelines and examples. In addition, this site provides directions on how to create a family pedigree.

Supplemental Documents

Title: Genetics Terminology Word List

File Name: [Sex linked disorders word list.doc](#)

Description: This document contains a list of genetic terminology that students should be familiar with before continuing this lesson.

Title: Definitions for Sex-linked Disorder Vocabulary

File Name: [Sex-linked word definitions.doc](#)

Description: This documents provides brief and easy to understand definitions to the vocabulary for sex-linked disorders.

Title: Pedigree Symbols

File Name: [Pedigree Symbols.doc](#)

Description: This Word document contains some commonly used pedigree symbols. Additional symbols can be found in the Web resources.

Step 2

Duration: 55 minutes

Notes on Sex-Linked Genes and Inheritance

In this step, students will learn about sex-linked genes, sex chromosomes and inheritance using the “Lecture Notes for Sex-Linked Disorders” provided as a supplemental document. This lecture will begin with a general discussion about genes, chromosomes, and inheritance and then will move on to sex-linked genes, followed by sex chromosomes. Please go through the lecture notes carefully and conclude by answering any questions about the material covered. The Web resources provide additional information on genetic counseling.

Web Resources

Title: Genetic counseling

URL: http://www.accessexcellence.org/AE/AEC/CC/counseling_background.html

Description: This National Health Museum Web site contains information on genetic counseling.

Title: Frequently Asked Questions about Genetic Counselors and the National Society of Genetic Counselors

URL: http://www.nsgc.org/consumer/faq_consumers.asp

Description: This National Society of Genetic Counselors Web site contains frequently asked questions and answers about genetic counseling.

Supplemental Document

Title: Lecture Notes for Sex-Linked Disorders

File Name: [Lecture Notes.doc](#)

Description: These lecture notes discuss the genetics of sex-linked genes and sex chromosomes.

Step 3

Duration: 15 minutes

Pedigree Practice

Using what the students have just learned from the lecture notes, they will answer questions in the “Pedigree Practice Sheet” included as a supplemental document. The students should work in groups or individually, but should be brought back together for discussion of the pedigree charts when they have completed them. Use the “Pedigree Practice Answer Key” to check for accuracy and to facilitate this discussion.

Supplemental Documents

Title: Pedigree Practice Sheet

File Name: [Pedigree Practice Sheet.doc](#)

Description: Word problems that allow the students to create three different pedigrees and answer a question about each of them.

Title: Pedigree Practice Answer Key

File Name: [Pedigree Practice Answer Key.doc](#)

Description: Answer key with pedigrees drawn out and answers to the questions posed on the pedigree practice sheet.

Step 4

Duration: 20 minutes

Discussion and Summarization

After the students have completed their pedigrees, have them review additional concepts through the “What Disorder Am I?” game. In this game the students will divide into teams and quiz each other on important concepts and disorders discussed in the lecture notes. The directions are included in the “What Disorder Am I?” document and the cards for the game are included in the “Cards for ‘What Disorder Am I?’ Game” document. After the students have finished the game, bring the students back together as a group and ask them to describe some types and characteristics of muscular dystrophy. This discussion will lead them into the research they will be conducting on muscular dystrophy in the following step.

Supplemental Documents

Title: What Disorder Am I?

File Name: [Lecture Review Game - Directions.doc](#)

Description: This document includes directions for a game to review the information presented in the “Lecture Notes for Sex-Linked Disorders” in Step 2. In addition, the game serves as a lead in to the muscular dystrophy research students will conduct in Step 5.

Title: “Cards for ‘What Disorder Am I?’ Game”

File Name: [Cards for Lecture Notes Game.doc](#)

Description: This document contains game cards for the “What Disorder Am I?” game.

Step 5 minutes

Duration: 1 hour, 20

Research and Presentation

Group the students into groups of two. Assign the group a research topic, one of the types of muscular dystrophy from the “Lecture Notes for Sex-Linked Disorders” in Step 2. Ask the students to break up the questions listed in the “Muscular Dystrophy Research Guidelines” included as a supplemental document in this step. An example of answers to these questions is provided in the “Research Answer Key”. Once all of the questions have been researched and answered, create a story board. The storyboards need to be on either a tri-fold board or a piece of poster board. See the “Display Board Rubric” for the information the storyboard should contain. After all students have finished their boards, have the students present their information to the class.

Web Resources

Title: Muscular Dystrophy Association Web site

URL: <http://www.mdausa.org/>

Description: This Muscular Dystrophy Association (MDA) Web site contains links to the different types/forms of the muscular dystrophy.

Title: Becker Muscular Dystrophy

URL: <http://www.mdausa.org/disease/bmd.cfm>

Description: This MDA Web site provides information on Becker muscular dystrophy. In addition, this site contains links that students should be encouraged to explore.

Title: Ducheene Muscular Dystrophy Web Site

URL: <http://www.mdausa.org/disease/dmd.cfm>

Description: This MDA Web site provides information on Duchenne muscular dystrophy. In addition, this site contains links that students should be encouraged to explore.

Title: Emery-Dreifuss Muscular Dystrophy Web Site

URL: <http://www.mdausa.org/disease/edmd.cfm>

Description: This MDA Web site provides information on Emery-Dreifuss muscular dystrophy. In addition, this site contains links that students should be encouraged to explore.

Title: Muscular Dystrophy Association Diseases Page

URL: <http://www.mdausa.org/disease/index.html>

Description: This Web site is the start of MDA’s diseases page. It contains links to many of the types of muscular dystrophy with information that could be included on the poster boards.

Title: Facts About Duchenne and Becker Muscular Dystrophies

URL: <http://www.mdausa.org/publications/fa-dmdbmd-what.html>

Description: This MDA Web site contains information on Duchenne and Becker muscular dystrophies, two common types.

Title: National Institute of Neurological Disorder and Stroke’s Muscular Dystrophy Information Page

URL: <http://www.ninds.nih.gov/disorders/md/md.htm>

Description: This National Institutes of Health Web site contains basic information on muscular dystrophy. Also, it includes information on several of the different types of muscular dystrophy.

Supplemental Documents

Title: Muscular Dystrophy Research Guidelines

File Name: [Muscular Dystrophy Research Guidelines.doc](#)

Description: These research guidelines provide a set of questions for students to answer while doing their research on their type of muscular dystrophy.

Title: Research Answer Key

File Name: [Muscular Dystrophy Research Guidelines Answer Key.doc](#)

Description: This document includes possible answers to the “Muscular Dystrophy Research Guidelines” questions for Duchenne muscular dystrophy.

Title: Display Board Rubric

File Name: [SexLinked ChromDisorder- Rubric.doc](#)

Description: This rubric provides guidelines for scoring storyboard project. It can be used for student self-assessment as well as teacher assessment.

Assessment

Students will be assessed on their creation of pedigrees in Step 3 using the “Pedigree Practice Answer Key” and on their generation of a sex-linked disorder display board using the “Display Board Rubric” in Step 5.

Modifications

Extension

Further Internet research on other types of sex-linked disorders with interested students could be facilitated. Additionally, a guest speaker from a local support group or a special education teacher could speak to the class about disability and health.

Web Resources

Title: Genetic Pedigrees

URL: <http://www.msu.edu/~langley6/chs/Bio/Genetics/Peddirect.htm>

Description: This is a Web site that students can use to generate family pedigree. It provides a great for extra credit.

Education Standards

National Science Education Standards

LIFE SCIENCE, CONTENT STANDARD C:

As a result of their activities in grades 9-12, all students should develop understanding of

- The cell
- **Molecular basis of heredity**
- Biological evolution
- Interdependence of organisms
- Matter, energy, and organization in living systems
- Behavior of organisms

SCIENCE IN PERSONAL AND SOCIAL PERSPECTIVES, CONTENT STANDARD F:

As a result of activities in grades 9-12, all students should develop understanding of

- Personal and community health
- Population growth
- Natural resources
- Environmental quality
- Natural and human-induced hazards
- **Science and technology in local, national, and global challenges**

Georgia State Science Standards

Grade: 9-12, Science, Applied Biology/Chemistry II 8

Topic: Continuity of Life

Standard: Explains what animal breeders need to know about genetic inheritance in animals to produce more economically valuable breeds.

Grade: 9-12, Science, Biology 10

Topic: Genetics (Mendelian Genetics)

Standard: Explains and uses the basic Mendelian genetic principles.

Sex-Linked Disorders Word List

Sex-Linked Chromosomal Disorders
April Jones, CDC's 2004 Science Ambassador Program

1. Sex linked
2. Chromosomes
3. Autosome/autosomal
4. Pedigree
5. Recessive
6. Dominant
7. Gene
8. X chromosome
9. Y chromosome
10. Sex chromosomes
11. Genotype
12. Phenotype
13. Heredity
14. Alleles
15. Traits
16. Centromere
17. Homozygous
18. Heterozygous
19. Genetic disorder
20. Mutation

Word list came in part from:

Biology: The Dynamics of Life, McGraw-Hill Company, Inc., 2002, Columbus, OH.

Sex-Linked Disorder Definitions

Sex-Linked Chromosomal Disorders
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1. Sex linked traits -- traits that are controlled by genes located on the sex chromosomes.
2. Chromosomes – cell structure that carries the genetic material that is copied and passed from one generation to the next.
3. Autosome/autosomal – pairs of matching homologous chromosomes in somatic cells.
4. Pedigree – graphic representation of genetic inheritance used to map genetic traits.
5. Recessive – trait of an organism that can be masked by the dominant form of a trait.
6. Dominant – observed trait of an organism that masks the recessive form of a trait.
7. Gene – segment of DNA that controls the protein production and the cell cycle.
8. X chromosome – one of the sex chromosomes
9. Y chromosome – one of the sex chromosomes, responsible for determining the sex of a gamete.
10. Sex chromosomes – in humans, the 23rd pair of chromosomes; determine the sex of an individual and carry sex-linked characteristics. Containing the X and Y chromosomes.
11. Genotype -- combination of genes in an organism.
12. Phenotype – outward appearance of an organism, regardless of its genes.
13. Heredity – passing on of characteristics from parents to offspring.
14. Alleles – alternative forms of a gene for each variation of a trait of an organism.
15. Traits – characteristic that is inherited; can be either dominant or recessive.
16. Centromere – cell structure that joins two sister chromatids of a chromosome.
17. Homozygous – paired chromosomes with genes for the same traits arranged in the same order.
18. Heterozygous – two different alleles for a trait
19. Genetic disorder – disorder related to the genes or chromosomes.
20. Mutation – any change or random error in a DNA sequence.

Reference

Biology: The Dynamics of Life, McGraw-Hill Company, Inc., 2002, Columbus, OH.

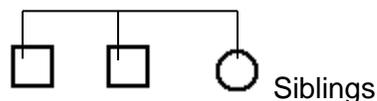
Pedigree Symbols

Sex-Linked Chromosomal Disorders
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Symbol	Description
	Unaffected male
	Unaffected female
	Affected male
	Affected female
	Deceased male
	Deceased female
	Possibly affected male or unknown
	Possibly affected female or unknown
	Carrier male of autosomal recessive disorder
	Carrier female of autosomal recessive disorder
	Carrier female with an X-linked disorder

Sometimes these symbols will be used in families with 2 different disorders – each half will tell whether the person is affected for that disease.

Connected Symbols



Source:

Coriell Institute for Medical Research. Pedigree Symbol Definitions [online]. 1998. [cited 2004 July 16]. Available from URL: <http://locus.umdj.edu/ada/gennid/phase1/pedigrees/pedhelp.html>.

Lecture Notes about Sex-Linked Genes

Sex-Linked Chromosomal Disorders
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General Genetic Information

Autosome

All chromosomes (1-22) except the sex chromosomes

Sex chromosome

The Y and X chromosomes

Sex determination

- autosomes and sex chromosomes segregate during meiosis (review meiosis if necessary)
 - sex of offspring is determined by the combination of sex chromosomes in the fertilized egg. The mother can only contribute an X chromosome because her genotype is XX, however the father can contribute either an X or a Y, because his genotype is XY. Therefore, the genotype of the off-spring is either male or female depending on whether the gamete receives an X or Y from the father.
 - o Most organisms: XX=female XY=male
 - The sex of some animals is determined in different ways.
 - o Grasshoppers XX=girl XO=male
 - o Ants and bees have NO sex chromosomes—sex determined by total chromosome number.
 - Males-unfertilized egg, haploid Females-fertilized egg, diploid
 - o Fish and reptiles-sex determined by factors in the environment—like the temperature
-

Sex-Linked Genes

Sex-linked gene

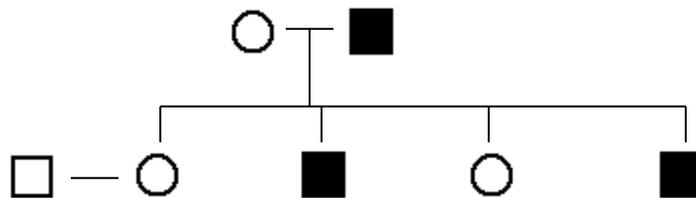
A gene that is located on the X or Y chromosomes

Sex-linked gene examples:

- White-eyes in fruit flies: If a fruit fly is white-eyed, then it must be male because the gene that makes the color of the eyes white is located on the Y chromosome. White eyes in fruit flies is a Y-linked trait..
- Muscular dystrophy: Characterized by progressive weakness and degeneration of the skeletal and voluntary muscles with control movement
 - a. Affects people of all ages
 - b. Different forms have different onset ages, ranges from infancy to middle age or later
 - c. No specific treatment for any of the forms
 - d. Life expectancy of individuals varies by form
 - e. Examples of types of muscular dystrophy include:
 - i. Duchenne Muscular Dystrophy, with an onset of 2 to 4 years, (X-linked)

- ii. Becker Muscular Dystrophy, with an onset in the teen and young adult years, (X-linked)
 - iii. Emery-Dreifuss Muscular Dystrophy, with an onset of childhood or adulthood, (X-linked or autosomal)
 - iv. Note: there are other forms of muscular dystrophy that are autosomal instead of X-linked
- Hairy ears in humans: The gene for hairy ears is found only on the Y chromosome (Y-linked). Therefore, males are the only ones to have this sex-linked gene.

Below is an example of a pedigree for a Y-linked gene.



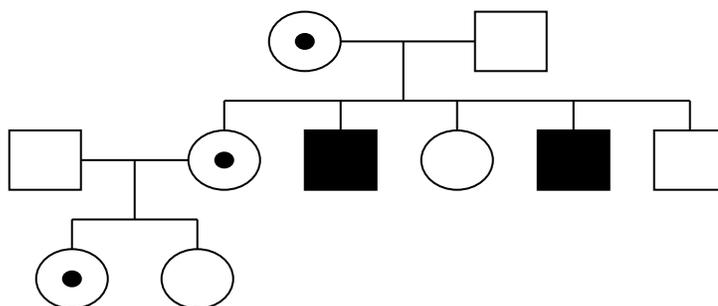
- Color-blindness in humans: The gene for color blindness is located on the X chromosomes (X-linked) and is recessive. This means that color blindness affects more males than females because males only get one X chromosome while females get two X chromosomes. If a female has the gene for color blindness on one of her X chromosomes, it is likely that she will have the dominant gene on her other X chromosome and therefore will not express the trait. On the other, if a male has the gene for color blindness he does not have a second gene that could be dominant, so he will express the trait.
- Hemophilia in humans: Like color-blindness, the gene for hemophilia is recessive and located on the X chromosome (X-linked). This means that males are more likely to be affected than females as explained above.

Historically, hemophilia has had an interesting effect on Queen Victoria and her family. She possessed the X-linked gene for hemophilia and passed this gene on to her children. This gene was passed down the royal family line, eventually affecting the youngest son of Tsar Nicolas the II, who received the gene from his mother, the granddaughter of Queen Victoria.

Reference

Huskey, Robert J. A Pedigree of Hemophilia in the Royal Families of Europe [online]. 1998. [cited 2004 June 30]. Available from URL: <http://www.people.virginia.edu/~rjh9u/roylhema.html>.

Pedigree for an X-Linked Gene



In the above pedigree, the mother is a carrier who passed the disorder to two of her sons. She has also passed on the gene to her one daughter, making that daughter a carrier. This daughter then passed the gene on to one of her own daughters; therefore, she also became a carrier.

Genetic Counseling

What is genetic counseling?

Genetic counseling is effort of genetic counselors, doctors and healthcare providers to help people with genetic disorders. Genetic counseling can include the following:

- an assessment of the patient's heritable risk factors
- information concerning the consequences of the disorder
- information on the probability of developing or transmitting the disorder,
- a discussion of ways in which the genetic disorder can be prevented, treated, or managed
- assistance in dealing with psychosocial issues (e.g. guilt, blame, shame)

Who is involved in genetic counseling?

Genetic counseling can be provided to those directly affected and their family members. Genetic counseling can be performed by many different healthcare professionals including doctors, nurses, therapists, scientists or psychologists. The title "genetic counselor" specifically refers to someone with a master's degree in the field. Others might do genetic counseling, but they are not referred to as genetic counselors.

Who receives genetic counseling?

The whole family of a patient with a genetic disorder receives genetic counseling; however, a person may receive genetic counseling if he/she knows a disorder runs in their family before they decide to have children. It can also be very rewarding to finally understand inheritance patterns.

What is the process of genetic counseling like for the recipients?

The process of genetic counseling can be very difficult for the recipients. They are often asked very personal questions and often required to have medical testing or medical records available to determine if a particular gene is present.

What are the techniques used in genetic counseling?

Genetic counselors use pedigrees to determine family history and inheritance patterns, medical tests like karyotyping or DNA analysis, X-rays, ultrasounds, urine analysis, skin biopsy, or physical exams.

What are the biggest challenge for genetic counselors?

The biggest challenge of genetic counseling is helping families cope with the emotional, psychological, medical, social and economic consequences of genetic disease.

How do patients react during genetic counseling?

Patients react in different ways when they learn their genetic risk status. Some take the information matter-of-factly. Others react with anger, shock, denial, grief, depression, confusion, and guilt.

What are some positive results from genetic counseling?

Genetic counseling may help patients decide if having children is an option and if a prenatal diagnosis is available. It may also help patients by letting them know that they are not alone and that help is available.

What are some of the adverse effects from genetic counseling?

Someone diagnosed with a genetic disease may be avoided by other relatives because the relatives do not know what to say or because they do not want to face up to the possibility that they too may develop the same genetic disease.

References

Resta, Robert G. Genetic Counseling: Coping with the Human Impact of Genetic Disease [online]. [Date Cited 10 October 2004] Available at URL: http://www.accessexcellence.org/AE/AEC/CC/counseling_background.html.

National Society of Genetic Counselors. FAQs about Genetic Counselors and the NSGC [online]. 1995-2004. [Date Cited 19 November 2004] Available at URL: http://www.nsgc.org/consumer/fag_consumers.asp.

Pedigree Practice Sheet

Sex-Linked Chromosomal Disorders
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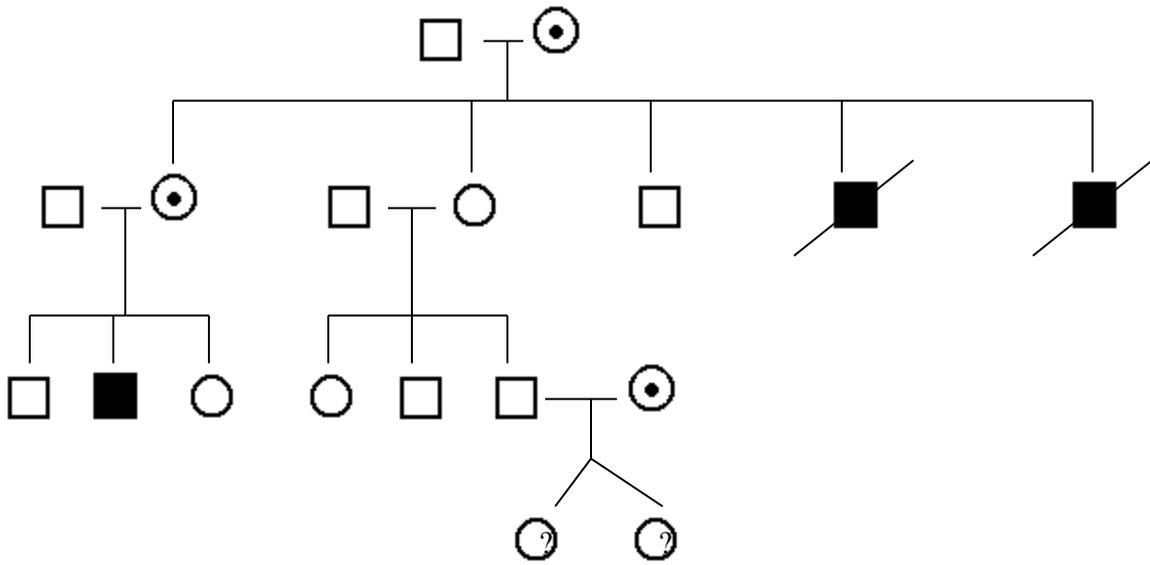
Use the information provided below to create a pedigree. Then answer the question at the end of each description.

1. A man and woman marry. They have five children, 2 girls and 3 boys. The mother is a carrier of hemophilia, an X-linked disorder. She passes the gene on to two of the boys who died in childhood and one of the daughters is also a carrier. Both daughters marry men without hemophilia and have 3 children (2 boys and a girl). The carrier daughter has one son with hemophilia. One of the non-carrier daughter's sons marries a woman who is a carrier and they have twin daughters. What is the percent chance that each daughter will also be a carrier?
2. The great-great maternal grandmother of a boy was a carrier for color-blindness, an X-linked disorder. His great uncle on his mother's side was colorblind but this great uncle's father was unaffected. The boy's mother has 2 brothers (1 colorblind, 1 unaffected) and 1 sister (unaffected). The boy's grandmother on his mother's side had 1 brother who was colorblind and 3 sisters. Two of these sisters were unaffected and one was a carrier. The boy's great grandmother on his mother's side had 4 sisters. The boy has one unaffected sister and he is colorblind. What is the probability of the boy's sons being colorblind if he marries a non-carrier?
3. An unaffected man marries a woman who is a carrier for Duchenne Muscular Dystrophy, which is attributed to an X-linked gene. They have four children, one with Duchenne, one carrier daughter and a daughter and son who are unaffected. The child with Duchenne Muscular Dystrophy dies in childhood. The carrier daughter marries and has three children of her own, two of which are carriers and one of which is unaffected. What is the most likely sex of these two carrier children given the fact that they are unaffected by the X-linked gene?

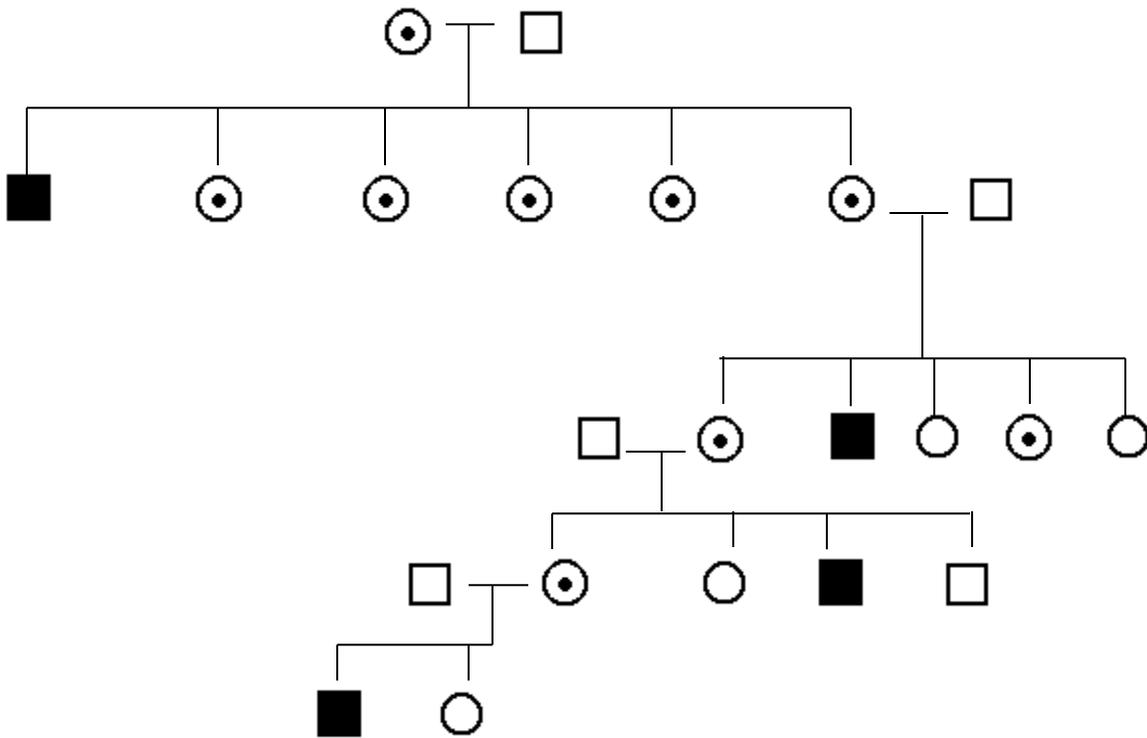
Pedigree Practice Answer Key

Sex-Linked Chromosomal Disorders
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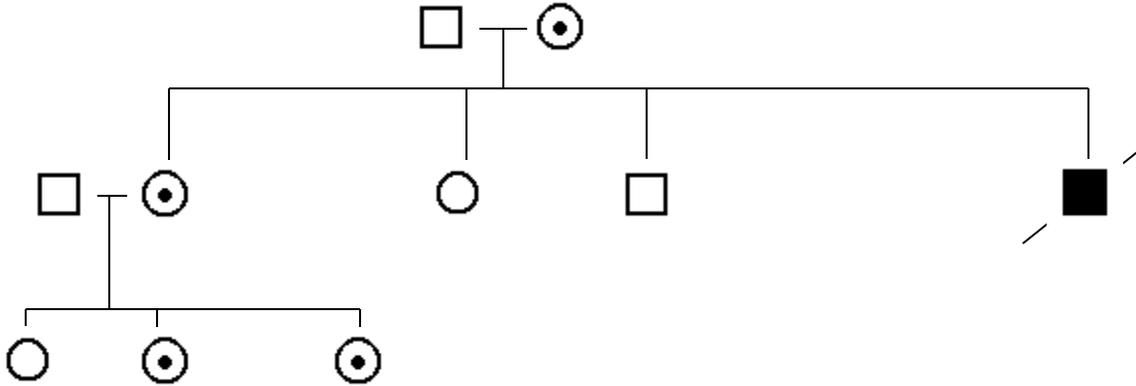
1. There is a 50% chance that each daughter will be a carrier. This is because the mother will contribute one X-chromosome to her daughter and there is a 50% chance that she will contribute the X chromosome that carries the gene as well as a 50% that she will contribute the X chromosome that does not carry the gene.



2. The probability that the boy's son will be colorblind is 0. This is because the gene is X-linked and he will contribute only his Y chromosome, not his X chromosome, to his son.



3. The two carrier children will be daughters. See pedigree chart below.



What Disorder Am I?

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Preparation

On different pieces of colored construction paper list the name of a disorder or concept from the lecture on one side of the paper and the definition any descriptive characteristics on the other side. Sample cards are provided as an attachment in step 4.

Directions

This activity can be done as a class or in small groups. Divide the class or small groups into two teams. Have a member from one team read the definition and any descriptive characteristics of a disorder or concept from one of the colored cards. If the opposing team is able to name the correct concept or disorder that matches the definition and descriptive characteristics, that team earns one point. If the team is unable to answer, then the team receives no points. The teams should take turns reading the cards and answering.

After the students have played for a while, have the asking team read the name of the disorder or concept and the answering team identify the definition or descriptive characteristics.

A Twist

Have the students pretend they are a patient in a doctor's office or that they are a genetic counselor. Then the patient from one team can ask about a disorder or concept and the genetic counselor from the other team can discuss the definition or descriptive characteristics keeping in mind the role of a genetic counselor.

Cards for “What Disorder Am I?” Game

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Please print the cards below. Cut out the cards along the solid line. Once cut the cards can be folded and glued or taped together so that the concept or disorder appears on one side of the card and the definition and characteristics appear on the other side.

Sex Chromosomes	I am your X and Y chromosomes. What am I?
Autosomes	I am all of your chromosomes except your sex chromosomes. What am I?
Emery-Dreifess Muscular Dystrophy	I degenerate skeletal and voluntary muscles. I am caused by an X-linked or autosomal gene and affect people between their childhood and adulthood. What am I?

Color Blindness	I might cause you not to see green or blue colors. I am an X-linked condition. What am I?
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Duchenne Muscular Dystrophy	I degenerate skeletal and voluntary muscles. I am caused by an X-linked gene and affect people between the ages of two and four. What am I?
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Becker Muscular Dystrophy	I degenerate skeletal and voluntary muscles. I am caused by an X-linked gene and affect people between their teen years and young adulthood. What am I?
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<p>Sex Determination</p>	<p>I am a concept that associates two X chromosomes with a female and one X and Y chromosomes with a male in humans. What am I?</p>
<p>Hemophilia</p>	<p>I am an X-linked condition that could cause you to bleed to death from a small razor nick. What am I?</p>
<p>Queen Victoria</p>	<p>I am a famous queen who passed on the X-linked gene for hemophilia to my children in the royal family. Who am I?</p>

Muscular Dystrophy	I cause progressive weakness and the degeneration of skeletal and voluntary muscles. I have many different types. What am I?
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Sex-Linked Gene	I am a gene that is located on the X or Y chromosome. What am I?
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Genetic Counseling	I might include an assessment of a patient's heritable risk factors as well as information on the probability of developing or passing on particular disorders. What am I?
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Muscular Dystrophy Research Guidelines

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Type of Muscular Dystrophy: _____

Group Members: _____

1. Who discovered the type of muscular dystrophy you are researching?
2. Why is this type named the way it is?
3. What causes it?
4. What are the characteristics of this type?
5. What age group does it affect?
6. Are there any treatments available?
7. What tests are used to diagnose the disorder?
8. What current research is being done?
9. What is the prognosis?
10. Include a quote from a person with the disorder and picture if possible.
11. Include a quote from a scientist on the type.

Muscular Dystrophy Research Guidelines Answer Key

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Example Answers for Duchenne Muscular Dystrophy

1. Who discovered the type of muscular dystrophy you are researching?
Duchenne muscular dystrophy (DMD) was first described by the French neurologist Guillaume Benjamin Amand Duchenne in the 1860s. (1)
2. Why is the type named the way it is?
See above question.
3. What causes it?
The absence of dystrophin, a protein that helps keep muscle cells together causes Duchenne muscular dystrophy. (3)
4. What are the characteristics of this type?
DMD primarily affects boys and is the result of mutations in the gene that regulates dystrophin, a protein involved in maintaining the integrity of muscle fiber. This type is X-linked recessive. DMD primarily affects boys, who inherit the disease through their mothers. Women can be carriers of DMD but usually exhibit no symptoms. (1,2)
5. What age group does it effect?
The onset of Duchenne occurs between 3-5 years of age and progresses rapidly. Most boys become unable to walk at 12, and by 20 have to use a respirator to breathe. (2)
6. Are there any treatments available?
There is no specific treatment for any of the forms of MD. Respiratory therapy, physical therapy to prevent painful muscle contractures, orthopedic appliances used for support, and corrective orthopedic surgery may be needed to improve the quality of life in some cases. Corticosteroids such as prednisone can slow the rate of muscle deterioration in patients with DMD but causes side effects. (2)
7. What tests are used to diagnose the disorder?
A patient and family history, a physical examination, genetic testing like karyotyping, a muscle biopsy and occasionally, special tests called electromyography or nerve conduction studies are done. The availability of DNA diagnostic tests, using either blood cells or muscle cells to get precise genetic information, is expanding rapidly. (1)
8. What current research is being done?
MDA-supported researchers have created a working dystrophin gene without the DMD mutation, and they've shown that it can strengthen muscle. Human trials of *gene therapy* for DMD and BMD are on the near horizon Other teams of MDA scientists are using *stem cells* generated from muscle and bone marrow to regenerate some muscles in laboratory models of Duchenne muscular dystrophy. Research teams are also exploring ways to get the body to generate more *utrophin*, a protein that may be able to substitute for dystrophin. (1)

9. What is the prognosis?

DMD eventually affects all voluntary muscles, the heart, and breathing muscles. Survival is rare beyond the early 30s. (2)

10. Include a quote from a person with the disorder or a parent with a child with the disorder and picture if possible.

“When my husband, Terry, and I were told eight years ago that our eldest child, Michael, has Duchenne muscular dystrophy, we were devastated. At first, we felt quite helpless. But knowing that helplessness wouldn't help our son, we became involved with the Muscular Dystrophy Association and with other parents. Since then we've learned a great deal, much of it very hopeful. We've also found inner peace with the challenges facing us, and enjoyed a full, rewarding family life. As you face the coming years, just remember: MDA and all its resources are there to help you see that your child is as happy and healthy as possible. You're not alone.

I wish you all the strength, hope and support you need.”



**Suzan Norton
Standish, Maine (1)**

(Please have your students include a picture)

11. Include a quote from a scientist on this type.

“One challenge the dystrophin gene presents is its enormous size. The gene is the largest gene yet identified in humans. Most vectors (usually modified viruses) available for gene replacement cannot incorporate a gene of this magnitude. The size probably also contributes to the high rate of new mutations in the gene and to the large number of different mutations that can occur within the gene. Definitive therapy may require precise knowledge of the particular gene defect in each patient.”

Audrey S. Penn, M.D., Acting Director National Institute of Neurological Disorders and Stroke (4)

References

1. Muscular Dystrophy Association. Facts About Duchenne and Becker Muscular Dystrophies (DMD and BMD) [online]. May 2000. [cited 2004 Oct 7]. Available from URL: <http://www.mdausa.org/publications/fa-dmdbmd-what.html>.
2. The National Institute of Neurological Disorders and Stroke. NINDS Muscular Dystrophy (MD) Information Page [online]. 29 Sept. 2004. [cited 2004 Oct 7]. Available from URL: http://www.ninds.nih.gov/health_and_medical/disorders/md.htm.
3. Muscular Dystrophy Association. Duchenne Muscular Dystrophy (DMD). 2004. [cited 2004 Oct 7]. Available from URL: <http://www.mdausa.org/disease/dmd.cfm>.

4. The National Institute of Neurological Disorders and Stroke. Testimony on Muscular Dystrophy, February 27, 2001, Audrey S. Penn, M.D., Acting Director National Institute of Neurological Disorders and Stroke July 1, 2001. [cited 2004 Oct 26]. Available at URL: http://www.ninds.nih.gov/about_ninds/md_testimony.htm.

Sex Linked Chromosomal Disorder Rubric

Sex-Linked Chromosomal Disorders
April Jones, CDC's 2004 Science Ambassador Program

Student Name: _____ Class period: _____ Class name: _____

Points	50 – 43 pts	42 – 30 pts	29 – 5 pts
Content	Student answered 9- 10 questions on the research guidelines sheet with accuracy and used scientific language appropriately. **5 pts for each accurate & thorough answer. **4 pts may be given for an accurate, but less thorough answer.	Student answered 6-9 of the questions. Some answers lacked accuracy or thoroughness and/or appropriate scientific language.	Student answered 5 or fewer questions accurately and/or many answers lacked appropriate scientific language and thoroughness.
	5 – 4 pts	3 pts	2 – 1 pts
Creativity	Student included something unique and different in their presentation that made the audience pay more attention to the presentation.	Student's presentation was acceptable and displayed an average level of creativity.	Presentation materials and delivery did not demonstrate creativity or interest on the part of the student and did not hold the audience's attention.
	5 – 4 pts	3 pts	2 – 1 pts
Aesthetics-	Student used a variety of color to enhance the poster board. The colors were pleasing to the eye and appropriate for the presentation. The presentation material was eye-catching and not over crowded or too busy.	Color, fonts, and graphics were acceptable. Placement on the poster board was neat and contributed to overall appearance.	Use of color, graphics, text, and white space was not appropriate, or it detracted from the content of the presentation.
	20 – 15pts	14 – 6 pts	5 – 0 pts
Neatness	Student presentation was typed and placement on the poster board was appropriate. Information presented was legible from a distance.	Student presentation was hand written neatly and most of the information was legible from a distance.	Student presentation was sloppy. Little or no attempt was made to create a nice presentation. The information was not legible from a distance.
	20 – 15 pts	14 – 6 pts	5 – 0 pts
Presentation	Student spoke with authority on the subject matter. They presented the information in a professional manner and seemed generally interested in the topic. The student could answer questions presented to him/her about the subject.	Student knew some of the information well and they were somewhat interested in the topic. They could only answer a few questions presented by the audience.	Students knew very little about their topic. They were not interested in their topic and could answer one or fewer questions presented to them.

Student self-assessment: /100

Teacher assessment: /100

Comments:

