






Genetic Disease Case Studies

Abstract



A role-play scenario as a geneticist and genetic counselor exploring different diseases and their inheritance patterns. Students test out a career while practicing the basics of genetics. Mock medical test results and some medical texts are provided. Links to more reference materials provided.

Learning Objectives

-  Distinguish between dominant and recessive traits
-  Differentiate between phenotype and genotype
-  Set up and complete a monohybrid cross (Punnett square)
-  Interpret karyotype charts
-  Research diseases, interpret medical information and relay that information in a compassionate, professional manner

Logistics

Time Required

-  Class time: 80 - 120 minutes
-  Prep time: 20 - 60 minutes

Prior Viewing

-  Introduction to Punnett Squares video
<https://www.youtube.com/watch?v=MRo6jRnhZs>
-  Geneticist career video
<http://www.careergirls.org/careers/geneticist-1>
-  Introduction to Mendel's research (TedEd)
<http://www.youtube.com/watch?v=Mehz71CxjSE#t=39>

Materials

Student handouts, manila folders to hold the 'confidential' medical test results

Appropriate For

7 - 9th grade students

Student Instructions

1. Read through the instructions to understand your job and what your tasks for the day will be.
2. Write "Dr." in front of your name.
3. When you record your responses, write them exactly as you would say them to your patient. Short answers like "no" can be misinterpreted as uncaring, so use full sentences.
4. Remember that medical information is confidential, so keep it safely tucked in the confidential file.
5. You have loads of medical texts available to you to check your facts and brush up on your knowledge of medical conditions.
6. Even doctors consult experts or other doctors for second opinions. Ask those around you for their opinions if you are unsure.

Teacher Recommendations

This is a role-play activity where students pretend to be doctors and give genetic counsel to their patients. Have students call each other “Dr. so-and-so” as they consult each other or you to keep the professionalism high as they record their responses. Remind them that real doctors do consult other doctors and experts (you are the expert) on occasion, so they are allowed to talk to their classmates. As they work through their cases, they should consult the confidential medical records and medical texts to inform their answers. This lesson gives you the chance to walk around and help any struggling students one-on-one as they learn to interpret word problems and complete the Punnett squares. Some students will work much faster than others and they can become experts that can consult with the other doctors in the room once you’ve checked their work.

Assessment

The format of this lesson allows you to constantly work with students individually to assess their progress and teach or re-teach concepts. Tutoring is the most effective form of teaching and you are able to essentially tutor any student who needs it while the class is working. Here are some tips for effective ongoing assessment in this lesson:

- ⇒ Walk around the room and look to see if students have correct conclusions (but do it without disrupting their flow if possible)
- ⇒ If you see a mistake, instead of saying “number three is wrong” try asking a question like, “why did you make John homozygous dominant?” and have them guide you through their reasoning. Then you can lead them to uncover the correct answer for themselves. This process is more likely to lead to long-term understanding and memory for the student.
- ⇒ Direct students to the medical text, and guide them through the materials if they are struggling to interpret the information. Often students just need help finding the information, and then interpretation isn’t a problem.
- ⇒ Remember, you are the expert doctor in the room, keep your conversation professional and they will mimic you in their responses.

Answer Key

The way in which students write their answers will vary, but look for professionalism. The answers given in the key are just examples of how the responses might be written. The genetic information, however, should be the same (eg. 25% of having the disease).

Genetic Diseases Case Studies

Name _____ Date _____ Per _____

Introduction:

This is a role-play activity where you imagine yourself in the future, after years of education, as a doctor and genetic counselor working at the local hospital. You have many patients to work with every day and you have to help them understand the science behind their genetics questions in a way that is not overly complicated. Today, you are fully booked with patients that have many questions and concerns. You will be able to give them information, facts and suggestions, but decisions are ultimately up to them (despite what you might desire). Answer their questions, refer to their medical charts, and your medical texts as needed. Remember to answer their questions with care, empathy and kindness.

8:00am Paul and Melinda Carraway

- 1 Paul has two cousins with cystic fibrosis and is worried that he might carry the gene. His wife Melinda knows she is a carrier because she was tested after her younger sister was diagnosed. Should Paul be tested to find out if he is a carrier for cystic fibrosis? (being a *carrier* means that person is heterozygous and *carries* the recessive allele)
- 2 What are the chances of Paul and Melinda having a child with cystic fibrosis if they are both carriers? (show a Punnett square)
- 3 If a child is born with CF, what struggles will they have in their lifetime?

9:00am Benjamin and Leah Cohen

- 1 Leah's brother (who died before she was born) had Tay-Sachs disease. She is worried that she is a carrier for the disease and has had a genetic test performed. She is here for the results. Look at her blood test and see if she is positive (has the gene) or negative. Is she a carrier?
- 2 Benjamin has no history of Tay-Sachs in his family, but he is of Jewish descent. Would you recommend that he also be tested?
- 3 If Benjamin does not have the gene, what is the likelihood that they would have a child with Tay-Sachs? (show a Punnett square)

4. What happens to a child born with Tay-Sachs?

10:00am Angela and Sal Smith-Cook

1. Angela is 42 and pregnant. On her last visit she had the fetus' amniotic fluid tested for chromosomal abnormalities. She really wants to have a girl, but only a healthy "normal" girl. She has decided she will not carry a baby with abnormalities to term. Check the karyotype and write what you will tell Angela and Sal about their baby.

11:00am John Remmin

1. John's father was diagnosed with Huntington's disease at 32 years old – only 2 years after John was born. Now that John is married and thinking about having children of his own, he is concerned that he might also have Huntington's disease. His test results are in, what will you tell John? (Hint: it is very rare to have two genes for Huntington's disease)

2. If John decides to have children, what are the chances that they will have Huntington's disease? (show punnett square)

3. What symptoms can John expect to experience in a few years?

1:00pm Keenan and Demetria Davis

1. Keenan and Demetria both grew up in the United States but their parents immigrated from South Africa. They want to start a family, but both of them had one parent with Sickle Cell Anemia. They have both been healthy their whole lives and have been tested to see if they carry the gene. Look at their blood work and tell them their results.

2. What are the chances that they will have a child with sickle cell anemia? (Show punnett square)

3. What are the chances that they will have a child who carries the gene?

4. What are the chances that they will have a child who does not have the gene at all?

2:00pm Amy and Matt Rolof.

1. Amy and Matt Rolof are both little people with Achondroplasia. They want to know if they have any chances of having children with normal height, children who are little and also, if they could have stillborn children. Use a Punnett square to explain their chances for all three phenotypes.

3:00pm Sandra and Matt Ellington.

1. Sandra is 25 and has hypochondriac tendencies. She constantly worries that she has many different diseases and that she will pass them all to her children. She is especially afraid that she will get breast cancer. Her mother, grandmother and older sister have all had breast cancer. She has been tested for the genes (BRCA1,2) and you are ready to give her the results. Choose your words carefully, and write them below.

2. What preventative measures could she take?

3. Is she likely to pass this on to her children? How do you know?

Medical Records

Medical Record for:

CONFIDENTIAL MATERIAL

Cohen, Leah Sex: F

234 Main St.
Washington

Date	Medical Test	Medical Test
Sept. 4	Blood Test Low hexosaminidase A activity Likely carries Tay-Sachs gene	

Medical Record for:

CONFIDENTIAL MATERIAL

Smith-Cook, Angela Sex: F

654 Aloha St.
Washington

Date	Medical Test	Medical Test
Sept. 2	Amniocentesis, fetus is 12 weeks	



Medical Record for:		CONFIDENTIAL MATERIAL
Remmin, John Sex: M 789 Wallow Way. Washington		
Date	Medical Test	Medical Test
Sept. 3	Blood Test 109 CAG repeats	

Medical Record for:		CONFIDENTIAL MATERIAL
Davis, Keenan Sex: M 567 E. 1 st Ave Washington		
Date	Medical Test	Medical Test
Sept. 4	Blood Test Low amounts of Hemoglobin S (HbSS)	

Medical Record for:		CONFIDENTIAL MATERIAL
Davis, Demetria Sex: F 567 E. 1 st Ave Washington		
Date	Medical Test	Medical Test
Sept. 4	Blood Test Low amounts of Hemoglobin S (HbSS)	

Medical Record for:		CONFIDENTIAL MATERIAL
Ellington, Sandra Sex: F 574 Blossom St. Washington		
Date	Medical Test	Medical Test
Sept. 4	Base pair analysis of BRCA1 45 mutations	Base pair analysis of BRCA2 20 mutations

Medical Texts

Tay Sachs

Healthy babies develop vision, movement, hearing, and other vital functions in part because enzymes clear out fatty protein and other unwanted material that can interfere with growth.

But a baby with Tay-Sachs disease is born without one of those important enzymes, Hexosaminidase A (Hex A). So, as those fatty proteins build up in the brain, they hurt the baby's sight, hearing, movement, and mental development. Children with the disease usually die before the age of five.

A child can only get Tay-Sachs by inheriting it. The genetic trait is relatively common among certain ethnic groups, such as Ashkenazi Jews. Tay-Sachs can be detected before birth, so couples in at-risk ethnic groups who are thinking of having children may want to get a blood test to find out whether their child would be likely to have it.

Who Is at Risk for Tay-Sachs?

Each year, about 16 cases of Tay-Sachs are diagnosed in the United States. Although Ashkenazi Jews (Jews of central and eastern European descent) are at the highest risk, it is now also prevalent in non-Jewish populations, including people of French-Canadian/Cajun heritage.

Some people carry the genetic mutation that causes Tay-Sachs, but do not develop the full-blown disease.

Among Ashkenazi Jews, 1 in 27 people are carriers; in the general population, 1 in 250 people are.

A child can only have Tay-Sachs disease if both parents are carriers of the gene.

Tay-Sachs is a recessive trait.

Achondroplasia dwarfism occurs as a sporadic mutation in approximately 85% of cases (associated with advanced paternal age) or may be inherited in an [autosomal dominant genetic disorder](#) that is a common cause of [dwarfism](#). A person with achondroplasia is heterozygous and has only one mutated gene (the other is normal). If both parents of a child have achondroplasia, and both parents pass on the mutant gene, then it is very unlikely that the [homozygous](#) child will live past a few months of its life (often the child is stillborn). It is possible for two parents with achondroplasia to pass on normal genes and have children of average adult heights. The disorder itself is caused by a change in the DNA for fibroblast growth factor receptor 3 which causes an abnormality of [cartilage](#) formation. Achondroplastic dwarfs have [short stature](#), with an average adult height of 131 cm (4 feet, 3½ inches) for males and 123 cm (4 feet, ½ inch) for females.

The prevalence is approximately 1 in 25,000.

Down Syndrome

Trisomy of the 21 chromosome.

Additional medical texts can be found at

<http://teach.genetics.utah.edu/content/health/ngs/NGS-student%20packet.pdf>

Genetic Diseases Case Studies

ANSWER KEY

Introduction:

This is a role-play activity where you imagine yourself in the future, after years of education, as a doctor and genetic counselor working at the local hospital. You have many patients to work with every day and you have to help them understand the science behind their genetics questions in a way that is not overly complicated. Today, you are fully booked with patients that have many questions and concerns. You will be able to give them information, facts and suggestions, but decisions are ultimately up to them (despite what you might desire). Answer their questions, refer to their medical charts, and your medical texts as needed. Remember to answer their questions with care, empathy and kindness.

8:00am Paul and Melinda Carraway

- 1 Paul has two cousins with cystic fibrosis and is worried that he might carry the gene. His wife Melinda knows she is a carrier because she was tested after her younger sister was diagnosed. Should Paul be tested to find out if he is a carrier for cystic fibrosis? (being a *carrier* means that person is heterozygous and *carries* the recessive allele)

Paul, considering the fact that you have two cousins with cystic fibrosis, and that this is a genetic disease, I highly recommend that you be tested for cystic fibrosis. Cystic fibrosis is a recessive gene so you may have the gene and not know it.

- 2 What are the chances of Paul and Melinda having a child with cystic fibrosis if they are both carriers? (show a Punnett square)

If you both are carriers for cystic fibrosis, there is a 25% chance that you will have a child with CF.

But, there is a 75% chance of having a child without the disease.

	F	f
F	FF	Ff
f	Ff	ff

- 3 If a child is born with CF, what struggles will they have in their lifetime?

CF causes poor digestion, dehydration, coughing and vomiting. As it progresses, teenagers have slowed growth, delayed puberty and less physical endurance. Adults may experience a collapsed lung, heart failure, infertility and frequent infections which can lead to death. Treatment costs an average of \$40,000 per year in the US.

9:00am Benjamin and Leah Cohen

- 1 Leah's brother (who died before she was born) had Tay-Sachs disease. She is worried that she is a carrier for the disease and has had a genetic test performed. She is here for the results. Look at her blood test and see if she is positive (has the gene) or negative. Is she a carrier?

Leah, your test results show a lower than normal Hexosaminidase -A activity, which means it's likely that you are a carrier for the recessive Tay-Sachs disease.

- 2 Benjamin has no history of Tay-Sachs in his family, but he is of Jewish descent. Would you recommend that he also be tested?

Benjamin, even though you don't have a personal family history of Tay-Sachs, I do recommend that you be tested for the gene because a carrier will show no physical traits and the gene can go unnoticed. This disease is highly prevalent in the Jewish community, so you do run a higher risk.

- 3 If Benjamin does not have the gene, what is the likelihood that they would have a child with Tay-Sachs? (show a Punnett square)

If you don't have the gene, Benjamin, then there is no chance that you will have a child with Tay-Sachs, even with Leah as a carrier.

	T	t
T	TT	Tt
T	TT	Tt

4. What happens to a child born with Tay-Sachs?

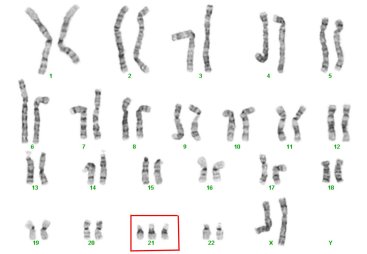
A baby born with Tay-Sachs will have fatty proteins build up in their brain which will hurt their sight, hearing, movement and mental capacity. Children with this disease usually die before the age of five.

10:00am Angela and Sal Smith-Cook

1. Angela is 42 and pregnant. On her last visit she had the fetus' amniotic fluid tested for chromosomal abnormalities. She really wants to have a girl, but only a healthy "normal" girl. She has decided she will not carry a baby with abnormalities to term. Check the karyotype and write what you will tell Angela and Sal about their baby.

Good morning Angela. We have the amniocentesis results here for you. I know that you were hoping for a girl, and the 23 chromosomes on the karyotype show two large X-chromosomes so you have a girl! Also, look at the spot here that says 21, there are three chromosomes where there are typically only two. This means that your daughter has Down Syndrome. I know you have a lot to think about now, and I encourage you to research and contact families who have a child with Down Syndrome to make an informed choice.

(teacher note: remind students that they cannot make the choice for their patients!)



11:00am John Remmin

1. John's father was diagnosed with Huntington's disease at 32 years old – only 2 years after John was born. Now that John is married and thinking about having children of his own, he is concerned that he might also have Huntington's disease. His test results are in, what will you tell John? (Hint: it is very rare to have two genes for Huntington's disease)

Hi John, we have your results here. We look for the number of CAG repeats in a certain section of your DNA to determine if you have Huntington's disease. Normally, there are between 10 and 35 repeats. More than that would indicate the Huntington's disease gene. You do have a much higher number of repeats, 109. It is good that you know about this now, so that you can make informed decisions for your future, and your family.

2. If John decides to have children, what are the chances that they will have Huntington's disease? (show punnett square)

John is probably heterozygous because it's unlikely to have two genes for the disease. His wife has no reported history of the disease so she has two normal genes. This disease is dominant so normal genes are recessive.

	H	h
h	Hh	hh
h	Hh	hh

There is a 50% chance that his children would have the disease.

3. What symptoms can John expect to experience in a few years?

In a few years you'll experience mild things like mood swings, depression, forgetfulness and lack of coordination. As it progresses, you'll have involuntary movements, memory declines and walking, speaking and swallowing ability will diminish. Eventually you will be unable to take care of yourself and death is usually from choking, infection or heart failure.

1:00pm Keenan and Demetria Davis

1. Keenan and Demetria both grew up in the United States but their parents immigrated from South Africa. They want to start a family, but both of them had one parent with Sickle Cell Anemia. They have both been healthy their whole lives and have been tested to see if they carry the gene. Look at their blood work and tell them their results.

I've got your test results here and both of you show a low amount of hemoglobin S (Hb SS) which are the mutated, or sickle shaped blood cells. This means that you both are in fact carriers for Sickle Cell Disease.

2. What are the chances that they will have a child with sickle cell anemia? (Show punnet square)

There is a 25% chance that you will have a child with sickle cell anemia.

	H	h
H	HH	Hh
h	Hh	hh

3. What are the chances that they will have a child who carries the gene?

There is a 50% chance that you will have a child who carries the gene, like you both do.

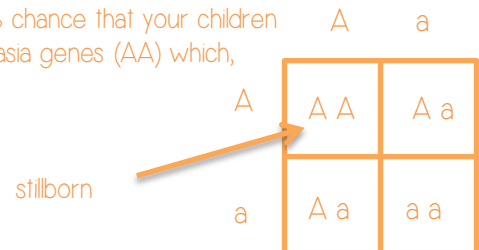
4. What are the chances that they will have a child who does not have the gene at all?

There is a 25% chance that your child will not have the gene at all

2:00pm Amy and Matt Rolof.

1. Amy and Matt Rolof are both little people with Achondroplasia. They want to know if they have any chances of having children with normal height, children who are little and also, if they could have stillborn children. Use a Punnett square to explain their chances for all three phenotypes.

Matt and Amy, because you are both heterozygous, this means that you have a 50% chance of having children who are also heterozygous (Aa) and have Achondroplasia. There is a 25% chance that your children will be of normal height, (aa) and a 25% chance that they will have two achondroplasia genes (AA) which, unfortunately, leads to early infant death or stillborn babies.



3:00pm Sandra and Matt Ellington

1. Sandra is 25 and has hypochondriac tendencies. She constantly worries that she has many different diseases and that she will pass them all to her children. She is especially afraid that she will get breast cancer. Her mother, grandmother and older sister have all had breast cancer. She has been tested for the genes (BRCA1,2) and you are ready to give her the results. Choose your words carefully, and write them below.

Hi Sandra, it's good to see you again. I have your results here today. What we look for is how many mutations you have in BRCA 1 and 2 because mutations indicate a higher risk of breast cancer. You have 45 mutations on BRCA 1 and 20 mutations on BRCA 2 which does indicate a higher chance of breast cancer. Again, you don't have breast cancer, just a higher risk. However, there are preventative things you can do.

2. What preventative measures could she take?

Eat a healthy diet and minimize your alcohol consumption, there are also some drugs that reduce the likelihood of the disease developing. Also, a very radical alternative is to have a double mastectomy which doesn't eliminate the possibility, but greatly reduces it.

3. Is she likely to pass this on to her children? How do you know?

This is an autosomal dominant gene, which means that if your children have just one BRCA 1 or 2 allele from you they have a higher risk of breast cancer. This doesn't mean that they will have cancer and it doesn't mean that you have cancer, just an increased risk.