To Hear – Genetic Deafness

Secrets of the Sequence Video Series on the Life Sciences • Grades 9 – 12

Teaching materials developed by VCU Life Sciences.

Virginia Commonwealth University

Classroom Tested Lesson

Video Description

"Secrets of the Sequence," Show 102, Episode 3

"To Hear-Genetic Deafness" – approximately 8 minutes viewing time

Maureen and Jim Hynes are the parents of two healthy, hearing children. This surprised them because both are deaf. Thanks to genetic testing, predicting deafness is easier. The numbers of deaf people aren't decreasing though – quite the opposite. Many deaf parents are choosing to have deaf children. This segment looks at the science and the controversy behind genetic deafness.

Ward Television
Producer: Paul Malkie
Associate Producer: Eric Wills
Featuring: Dr. Walter Nance, Human Genetics, Virginia Commonwealth University and Dr. Thomas Huff,
    Life Sciences, Virginia Commonwealth University
Lesson Author; Reviewers:Elizabeth Armstrong; Susan Walton and Peggy Deichstetter
Trial Testing Teacher: Nara Riplinger

National and State Science Standards of Learning

National Science Education Standards Connection

Content Standard A

As a result of activities in grades 9-12, all students should develop:
- abilities necessary to do scientific inquiry and
- be able to identify questions and concepts that guide scientific investigation.

Content Standard C

As a result of activities in grades 9-12, all students should develop an understanding of:
- molecular basis of inheritance.

Content Standard F

As a result of activities in grades 9-12, all students should develop an understanding of:
- personal and community health.

Content Standard G

As a result of activities in grades 9-12, all students should develop an understanding of:
- science as a human endeavor and
- the nature of scientific knowledge.

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 1 of 12

Selected State Science Standards Connection

Use http://www.eduhound.com (click on "Standards by State") or a search engine to access additional state science standards.

Virginia

BIO.1 The student will plan and conduct investigations in which:
- variables are defined and investigations are designed to test hypotheses;
- conclusions are formed based on recorded quantitative and qualitative data; and
- a scientific viewpoint is constructed and defended.

BIO.6 The student will investigate and understand common mechanisms of inheritance and protein synthesis. Key concepts include:
- prediction of inheritance of traits based on the Mendelian laws of heredity;
- uses, limitations, and misuses of genetic information; and
- exploration of the impact of DNA technologies.

North Carolina

Competency Goal 2: *The learner will develop an understanding of the continuity of life and the changes of organisms over time.*
Interpret and use the laws of probability to predict patterns of inheritance.
Assess the application of DNA technology to forensics, medicine, and agriculture.
Analyze and explain the role of genetics and environment in health and disease.
Examine the development of the Theory of Biological Evolution including:
- patterns
- variation
- natural Selection

Competency Goal 3: *The learner will develop an understanding of the unity and diversity of life.*
Determine the internal and external factors that influence the growth and development of organisms.

Overview

Given the genetic knowledge and counseling that are available for deaf parents, it would seem likely that there would be a continual decline in the number of deaf children born each year. However, the number of deaf people isn't decreasing – quite the opposite. Many deaf parents are choosing to have deaf children. This segment looks at the science and the controversy behind genetic deafness.

Thanks to genetic testing, predicting many forms of deafness is now easier. The number of genetic defects and the types of deafness they cause are greater than most people know. One genetic marker, referred to as the Connexion gene found on chromosome 13, interrupts the signaling between cells. Pendrid Syndrome is caused by an anomaly on chromosome 7, resulting in a malformation of the inner ear, as well as thyroid defects. Other genetic defects can cause malformation of the middle ear (Mondini Defect), absence of the middle ear (Michel Aplasia), or a defect of the cochlea or cochlear membrane (Scheibe Aplasia). One unique form of genetic deafness is Waardenburg's Syndrome; the carrier has eyes of two different colors and develops a white forelock. Deafness can be unilateral (one ear) - or bilateral (both ears), and can range from mild to profound.

Testing: A sample related multiple choice item from State Standardized Exams

Four o'clock flowers inherit flower color by incomplete dominance. If R represents the gene for red flowers and R′ represents the gene for white flowers, then what flower color phenotype would be the result of an RR′ x RR′ cross?

1. 25% red, 50% pink, 25% white*
2. 50% red, 25% pink, 25% white
3. 25% red, 25% pink, 50% white
4. 50% red, 50% white

(Source: North Carolina End-of-Course Test: Biology)

Video Preparation

Preview the video and make note of the locations at which you will later pause the video for discussion.

Before Viewing

1. Tell students about the following documented case of deafness in Massachusetts. In 1694 Jonathan Lambert arrived in a small town of Chilmark, Massachusetts. Jon was special because he was deaf. His condition was a genetic deafness that continued in the small community for generations. So many individuals inherited this condition that the basic means of communication in this community for both hearing and deaf persons became sign language. Being deaf in this community was not a disability. It was viewed only as a difference. Signing became a common part of day-to-day communication for citizens of Chilmark; much like being bi-lingual is today.

   Interested students may wish to read: Groce N.E _Everyone Here Spoke Sign Language:_ Heredity Deafness on Martha's Vineyard. 1985. Harvard University Press, Cambridge, or for more information about deafness in Chilmark visit http://deafness.about.com/cs/featurearticles/a/marthasvineyard.htm

2. Ask a student to look up the definition of "culture" and read it to the class:
   > _"The sum total of ways of living built up by a group of human beings and transmitted from one generation to another" Webster's New Universal Unabridged Dictionary, 1992.)_

   Then have students brainstorm the different cultures they are aware of in your local community. List these on the board.

3. Ask the following questions:
   - Can deafness be a culture?
   - If you think deafness is a culture, why do you think so?
     > _Deaf people have their own language, schools etc._
   - If you were deaf, would you choose to marry a deaf person? Why or why not?
     > _Note: this may bring up the subject of discrimination. Discuss the difference between making life choices and uneducated discrimination._
   - If you were deaf, would you choose to have a deaf child, if you could choose?" Why or why not?
   - Why do you think a deaf couple might prefer to have deaf children?

During Viewing

1. **START** the video.

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 3 of 12

2. **PAUSE** the video (3.40 minutes into the video) when the computer narrator says, "…there are an estimated 300 deafness genes, 70 have been mapped so far and more are being found each year."
   Ask:
   - "What are two physical causes for deafness?"
     malformation of the inner ear, and interruption of signal between the ear and the brain
   - Both Maureen and Jim are deaf, but their children are not. "Why aren't the children deaf?"
     Parents have different kinds of deafness
   - "If two people are deaf, would it be possible for them to determine the odds of having a deaf child?
     the parent's genotypes need to be determined by genetic testing. A Punnett square could be used to determine the probability of deafness-see Teacher notes/Background Information

3. **RESUME** the video.

4. **PAUSE** the video (5.42 minutes into the video) after the doctor says, "…a price we have to pay, as it were, for the freedom of association, for the freedom of being able to marry whomever you want to."

   Ask: "Why does science (and the general population) view deafness as a defect or handicap?"

5. **RESUME** the video and view to the end.

After Viewing

1. Lead a discussion using the following questions :
   - Should deaf parents be able to test their genes before having children?
   - What other factors should parents be able to screen for before they have children?
   - What factors shouldn't they be able to test for?
   - Should parents be able to test for certain factors present in a fetus?
   - Who should pay for genetic testing?

   Note: a trial test teacher discussed the fact that screening for the sex of the fetus is done in certain countries but this will lead to the moral issue that can cause over abortion. Since this is such a controversial topic, in the interest of time, you may want to limit the scope of the conversation.

2. Review the causes of genetic deafness.

3. Conduct the Student Activity: To Hear or Not to Hear

Teacher Notes for Student Activity; To Hear or Not to Hear

Introduction
Students will use the information given in the Student Handout to fill in the genotype and phenotype of the parents, complete the Punnett Squares and calculate the probability of deafness. In this activity, all genetic disorders are homozygous recessive. The first three are simple 2 X 2 Punnett squares but the last two are more challenging given the 4 X 4 permutations. If students have not studied Punnett Squares prior to this lesson, the information below should be reviewed.
Upon completion of this activity you may want, as an extension, to have the students play the role of genetic counselor. Each student will be assigned one of the families as "his/her case." They will find out more about the type of deafness and be able to play the role of genetic counselor by explaining to the "future parents" the probability of their child being deaf using their Punnett Square as part of their explanation, and how this type of deafness occurs.

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 4 of 12

Background Information:
Define the following terms for the students if they are unfamiliar with the vocabulary.

- Genotype--- *the genetic makeup of an individual*
- Phenotype--- *the observable or detectable characteristics of an individual organism*
- Homozygous--- *refers to a genotype consisting of two identical alleles of a gene for a particular trait.*
- Heterozygous--- *refers to a genotype consisting of two different alleles of a gene for a particular trait.*
- Recessive allele---*masked in the phenotype by the presence of a dominant allele*
- Dominant allele -- *masks the presence of a recessive allele in the phenotype*

Review how a Punnett Square in constructed in order to understand inheritance.

A.   You begin by drawing a grid of perpendicular lines:
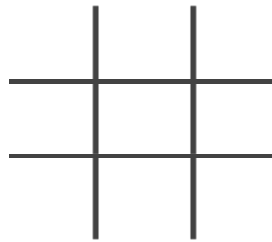
B: Next, you put the genotype of one parent across the top and that of the other parent down the left side. For example, if parent genotypes were YY and GG respectively, the setup would be:
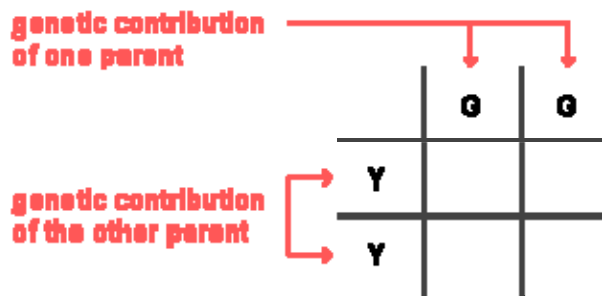

Note that only one letter goes in each box for the parents.   It does not matter which parent is on the side or the top of the Punnett square.

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 5 of 12

C.   Next, all you have to do is fill in the boxes by copying the row and column-head letters across or down into the empty squares. This gives the predicted frequency of all of the potential genotypes among the offspring each time reproduction occurs.
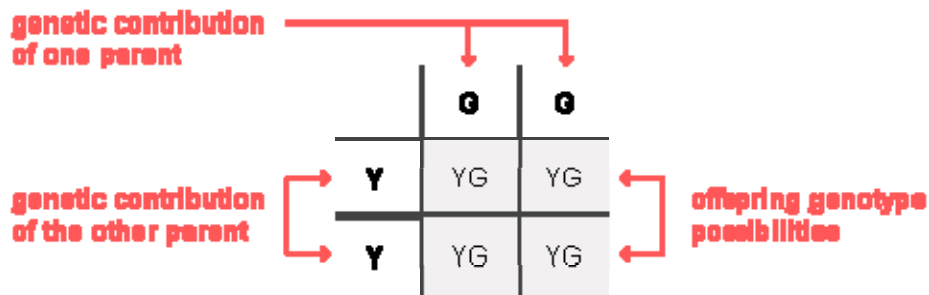
Results: In this example, 100% of the offspring will likely be heterozygous (YG). If the Y allele is dominant over the G allele, 100% of the YG offspring will have a Y phenotype, as Mendel observed in his breeding experiments.

In another example (shown below), if the parents both have heterozygous (YG) genotypes, there will be 25% YY, 50% YG, and 25% GG offspring on average. These percentages are determined based on the fact that each of the 4 offspring boxes in a Punnett square is 25% (1 out of 4). As to phenotypes, 75% will be Y and only 25% will be G. These will be the odds every time a new offspring is conceived by parents with YG genotypes.
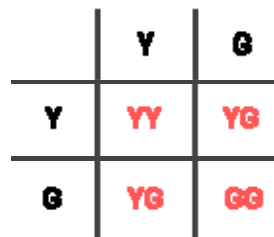
Conclusion: An offspring's genotype is the result of the combination of genes in the sex cells or gametes (sperm and ova) that came together in its conception. One sex cell came from each parent. Sex cells normally only have one copy of the gene for each trait (e.g., one copy of the Y or G form of the gene in the example above). Each of the two Punnett square boxes in which the parent genes for a trait are placed (across the top or on the left side) actually represents one of the two possible genotypes for a parent sex cell. Which of the two parental copies of a gene is inherited depends on which sex cell is inherited--it is a matter of chance. By placing each of the two copies in its own box has the effect of giving it a 50% chance of being inherited.

Answer Key: To Hear or Not to Hear

A: Couple One

Mother: deaf, Connexion
Father: heterozygous for Connexion
Genotype Mother ___cc_   Father___Cc__

Phenotype Mother_deaf__   Father_normal hearing_

Chance of having a deaf child

        ____50%

	c	c
C	Cc	Cc
c	cc	cc

B: Couple Two

Mother: carrier for Mondini Defect

Father: deaf, Mondini Defect

Genotype: Mother__Dd_   Father_dd__

Phenotype: Mother normal hearing Father_deaf

Chance of having a deaf child:

        50%

	D	d
d	d D	d d
d	d D	d d

C: Couple Three
Mother: deaf, Usher's Syndrome
Father: deaf, Usher's Syndrome

Genotype: Mother__uu___   Father__uu___

Phenotype: Mother _deaf__ Father_deaf__

Chance of having a deaf child:

	u	u
u	uu	uu
u	uu	uu

Courtesy of Virginia Commonwealth University · Richmond, Virginia · www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness · Page 7 of 12

_____100%

D: Couple Four

Mother: deaf, Michel Aplasia, carrier for Pendrid Syndrome

Father: deaf, Pendrid Syndrome, carrier for Michel Aplasia

Genotype M__mmPp___   F__Mmpp___

Phenotype M___deaf___   F__deaf____

Chance of having a deaf child __75%

	mP	mp	mP	mp
Mp	MmPp	Mmpp	MmPp	Mmpp
mp	mmPp	mmpp	mmPp	mmpp
Mp	MmPp	Mmpp	MmPp	Mmpp
mp	mmPp	mmpp	mmPp	mmpp

E: Couple Five

Mother: deaf, Connexion, carrier for Scheibe Aplasia
Father: deaf, Scheibe Aplasia, carrier for Connexion

Genotype: M__Sscc__   F__ssCc__

Phenotype: M__deaf__   F__deaf__

Chance of having a deaf child:

_____75%

	Sc	sc	Sc	sc
sC	SsCs	ssCc	SsCc	ssCc
sc	Sscc	sscc	Sscc	sscc
sC	SsCc	ssCc	SsCc	ssCc
sc	Sscc	sscc	Sscc	sscc

Student Handout: To Hear or Not to Hear

Introduction:

The value of studying genetics is in understanding how we can predict the likelihood of inheriting particular traits. One of the easiest ways to calculate the mathematical probability of inheriting a specific trait was invented by an early 20th century English geneticist named Reginald Punnett. His technique employs what we now call a Punnett square. This is a simple graphical way of discovering all of the potential combinations of genotypes that can occur in children, given the genotypes of their parents. It also shows us the odds of each of the offspring genotypes occurring.

For each of the following scenarios, complete a Punnett square to determine the genotype and phenotype. Express the chance of each couple having a deaf child as a percentage. Use the letters below to symbolize the disorder. Place the Mother and Father genotypes outside the box at the top and left side and the offspring genotypes in the inner boxes.

Note: all of these are recessive traits.

Connexion	(c)	Pendrid Syndrome	(p)
Mondini Defect	(d)	Usher's Syndrome	(u)
Michel Aplasia	(m)	Scheibe Aplasia	(s)

A: Couple One
Mother: deaf, Connexion
Father: heterozygous for Connexion

Genotype Mother _____   Father_____

Phenotype Mother_____   Father_____

Chance of having a deaf child

_____%

B: Couple Two
Mother: carrier for Mondini's Defect
Father: deaf, Mondini's Defect

Genotype: Mother_____   Father_____

Phenotype: Mother _____   Father_____

Chance of having a deaf child:

_____%

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 9 of 12

C: Couple Three
Mother: deaf, Usher's Syndrome
Father: deaf, Usher's Syndrome

Genotype: Mother_____   Father_____

Phenotype: M other_____ Father_____

Chance of having a deaf child:

_____%

D: Couple Four
Mother: deaf, Michel Aplasia, carrier for Pendrid Syndrome
Father: deaf, Pendrid Syndrome, carrier for Michel Aplasia

Genotype: Mother_____   Father_____

Phenotype: Mother_____   Father_____

Chance of having a deaf child

_____%

E: Couple Five

Mother: deaf, Connexion, carrier for Scheibe Aplasia
Father: deaf, Scheibe Aplasia, carrier for Connexion

Genotype: Mother_____   Father_____

Phenotype: Mother_____   Father_____

Chance of having a deaf child:

    _____%

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 11 of 12

Additional Resources

Because Web sites frequently change, some of these resources may no longer be available. Use a search engine and related key words to locate new Web sites

http://www.cdc.gov/ncbddd/ehdi/genetics.htm
The Center for Disease Control: The Genetics of Infant Hearing Loss.

http://www.healthwindows.org/body_genetics.asp
What is DNA? What are chromosomes? What are cells made of? What are genetic disorders and how are they caused? What do genetic counselors do? Can we cure diseases by fixing faulty genes?

www.kumc.edu/gec/resource.html

http://www.nidcd.nih.gov/health/hearing/index.asp
The site deals with; Hearing, Ear Infections, and the causes of Deafness or Hearing Loss, and Deafness Communication Options for People Who Are Deaf or Hard-of-Hearing

http://www.nidcd.nih.gov/news/releases/03/04_23_03.asp
Early diagnosis of usher syndrome type 1 made possible by new findings can help Ashkenazi Jewish children who are born deaf and have progressive loss of sight

http://ulysses.carthage.edu/faculty/msnavely/discov/genex.htm

http://www.kumc.edu/gec/resource.html

http://www.jbpub.com/genetics/geneticsweb.cfm

http://www.biology.arizona.edu/human_bio/problem_sets/ human_genetics/human_genetics.html

http://www.lsmsa.edu/MKhandoker/biology/Genetics/ Genetics%20laboratory%20exercises.htm

http://www.healthwindows.org/body_genetics.asp

http://www.kumc.edu/gec/support/hearing.html

http://www.ncbi.nlm.nih.gov/entrez/query

http://www.infanthearing.org/documents/HereditaryDeafness.pdf

http://www.jncasr.ac.in/anand/deafness.htm

Genomic Revolution
http://www.ornl.gov/sci/techresources/Human_Genome/education/education.shtmL
This Web site of the government-funded Human Genome Project has links about genomics, the history of the project, and more.

Secrets of the Sequence Videos and Lessons
This video and 49 others with their accompanying lessons are available *at no charge* from www.vcu.edu/lifesci/sosq

Courtesy of Virginia Commonwealth University • Richmond, Virginia • www.vcu.edu/lifesci
Made possible through the generous support of the National Academy of Sciences & the Pfizer Foundation
To Hear- Genetic Deafness • Page 12 of 12