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Bioinformatics and Genetic Testing

Introduction

Through a short topical play, students are introduced to the fields of **bioinformatics**, **genetic testing**, **direct-to-consumer genetic testing**, and ethical considerations. Students discuss some of the broad implications and ethical questions raised by gaining information through genetic testing. Students then consider a number of genetic tests and their potential usefulness and value and, as a class, explore the website of 23andMe, a company that offers direct-to-consumer genetic tests. The lesson wraps up as it began—by engaging students in a story. Through a short video, students are introduced to a family impacted by breast cancer. In *Lesson One*, students also learn how **bioengineers** might use bioinformatics tools in their career.

Learning Objectives

At the end of this lesson, students will know that:

- Genetic tests are available from commercial companies that market directly to consumers (direct-to-consumer genetic testing).
- Genetic tests are not available for all conditions or abilities, and vary in their usefulness and clinical validity.
- Genetic tests can have social and ethical implications.
- Bioinformatics tools are used by people in many career fields, including bioengineers.

At the end of this lesson, students will be able to:

- Recognize the social and ethical implications of genetic testing.
- Weigh some of the harms and benefits of direct-to-consumer genetic testing.
- Give examples of direct-to-consumer genetic tests.

Key Concepts

- The results of genetic tests can have social and ethical implications.
- A variety of genetic tests are available through companies that offer direct-to-consumer genetic testing, and the tests can vary greatly in their ability to predict disease.
- The field of bioinformatics uses computers to search biological databases, compare sequences, and represent protein structures.
- Bioinformatics gives us the tools to design and validate genetic tests.
- Bioinformatics tools are used by people in many careers, including bioengineers.

Class Time

Two class periods of 50 minutes each, 100 minutes total. One class period of 50 minutes may be sufficient if student discussion time is limited to 5–10 minutes for each activity, or if portions of the lesson are assigned as homework (see *Homework* section).

Prior Knowledge Needed

- An understanding of the Central Dogma of Molecular Biology (the flow of genetic information from DNA to RNA to protein).

Common Misconceptions

- A positive result from a genetic test means a person will certainly get that disease.
- Scientists and doctors understand the functions of all the genes in the human genome.
- Only some people have BRCA genes (i.e., people who have BRCA-related cancer).

Materials

Materials	Quantity
Copies of Student Handout— <i>Careers in the Spotlight</i>	1 per student
Copies of Student Handout— <i>Meet the Gene Machine</i>	2
Class set of Student Handout— <i>Direct-to-Consumer Genetic Testing Homework: 23andMe</i> <i>Optional: See Homework section</i>	1 per student (class set) (Optional)
Class set of Student Handout— <i>Understanding Genetics and SNPs</i> <i>Optional: See Teacher Resource—Direct-to-Consumer Genetic Testing Website Exploration</i>	1 per student (class set) (Optional)
Teacher Resource— <i>Discussion Questions for Meet the Gene Machine</i>	1
Teacher Resource— <i>Direct-to-Consumer Genetic Testing Website Exploration</i>	1
Teacher Answer Key— <i>Answers for Direct-to-Consumer Genetic Testing Homework: 23andMe</i> <i>Optional: See Homework section</i>	1 (Optional)
Teacher Answer Key— <i>Understanding Genetics and SNPs</i> <i>Optional: See Teacher Resource—Direct-to-Consumer Genetic Testing Website Exploration</i>	1 (Optional)
Cotton swab (prop for <i>Meet the Gene Machine</i>)	1
Large printout of paper either in a roll or sheet form (prop for <i>Meet the Gene Machine</i>)	1
A 'Gene Machine' (box or small lab machine) under a cover	1

Computer Equipment, Files, Software, and Media

Computer with internet access and projector to display PowerPoint slides, teacher-directed website exploration detailed in Teacher Resource—*Direct-to-Consumer Genetic Testing Website Exploration*, and the NOVA video segment, "A Family Disease."

Alternative: Print PowerPoint slides onto transparencies and display with overhead projector. The NOVA segment is available on DVD if internet access is not available.

Lesson One PowerPoint Slides—Bioinformatics and Genetic Testing.

Available for download at: <http://www.nwabr.org/curriculum/introductory-bioinformatics-genetic-testing>

A student version of lesson materials (minus teacher answer keys) is available from NWABR's Student Resource Center at: <http://www.nwabr.org/students/student-resource-center/instructional-materials/introductory-bioinformatics-genetic-testing>

Video: The eight-minute NOVA video segment "A Family Disease" from *Cracking the Code of Life* can be found streaming online at: http://www.pbs.org/wgbh/nova/genome/media/2809_q056_14.html. Clicking on the "Advanced Options" button leads to instructions for full-screen viewing. Purchasing information for the NOVA video can be found in the *Credit* section at the end of this lesson plan.

Teacher Preparation

- Load the classroom computer with the *Lesson One PowerPoint Slides* and the video segment "A Family Disease" (DVD or online streaming video).
- Make copies of the Student Handouts, one per student. Student Handout—*Careers in the Spotlight* will be used by students throughout the unit. Student Handout—*Direct-to-Consumer Genetic Testing Homework: 23andMe* is an optional homework assignment (see Homework section of this lesson plan), and is designed to be re-used as a class set.
- Set up two chairs at the front of the room, as for a talk show.
- Collect the props needed for the *Meet the Gene Machine* play.
- *Optional:* Assign two students or recruit two student or teacher volunteers to read the *Meet the Gene Machine* play before the in-class performance (for the roles of TV Talk Show Host and Scientist).

Procedure

Day 1

WARM UP

1. Explain to students the **aims of this lesson**. Some teachers may find it useful to write the aims on the board.
 - a. **Lesson Aim:** Introduce students to the field of bioinformatics.
 - b. **Lesson Aim:** Understand what type of genetic information a for-profit company (23andMe) is selling to the general public.
 - c. **Lesson Aim:** Introduce students to the *BRCA1* gene, which is involved in breast and ovarian cancer.

Teachers may also wish to discuss the *Learning Objectives* of the lesson, which are listed at the beginning of this lesson plan.

2. Tell students that the class will begin a unit of study about genetic testing and bioinformatics, and briefly define each term:

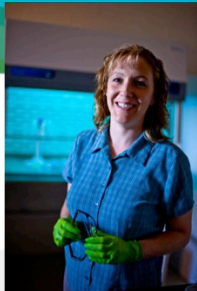
- **Genetic Testing** is the analysis of a person's DNA.

It is usually done to determine whether that individual carries changes (mutations) to genes that make them more susceptible to a disease. It is also popularly used to find out about ancestry and paternity.

- **Bioinformatics** is the application of computer science and information technology to biology and medicine.

Let students know that the focus of today's lesson will be on genetic testing, and that they will learn more about bioinformatics in *Lesson Two*. The topic of genetic testing can lead to some interesting social and ethical questions, as they will see in the play *Meet the Gene Machine*.

3. Show the PowerPoint for *Lesson One*, beginning with **Slide #1**. This slide highlights bioengineer Adrienne Minerick, PhD.



BIOENGINEER
ADRIENNE R. MINERICK, PhD

Place of Employment:
Michigan Technological University

Type of Research:
Biomedical Microdevices
For example, tiny devices to measure and manipulate living cells

"If a person is determined to learn, there will always be opportunities or resources for that person to pursue an education in science and engineering... There is a **real** demand for scientists and engineers whose contributions advance knowledge, technology, and the economic foundation of our society. I chose my career because I wanted to be a part of advancing knowledge and facilitating others to gain knowledge."

Bioinformatics & Genetic Testing: **Slide #1**

4. Give each student a copy of Student Handout—*Careers in the Spotlight*.
5. Students should think about, and write down, what kind of work a bioengineer might do (Bioengineer Question #1). This will be revisited at the end of the lesson.
6. Tell students to keep their *Careers in the Spotlight* handout available for future lessons.

PART I: Meet the Gene Machine

7. Introduce students to genetic testing and bioinformatics through a short play entitled *Meet the Gene Machine*. This comical play raises some of the broad implications of genetic testing.
8. Have the volunteer performers for the *Meet the Gene Machine* play come to the front of the room. Introduce one performer as Chris, a scientist, and the other as a popular TV talk show host.
9. Have students perform the play for the class.
10. After the play, use Teacher Resource—*Discussion Questions for Meet the Gene Machine* to lead a discussion about the play. Questions include:
 - What genetic traits are mentioned in the play?
 - Are genetic tests for all the traits equally valuable? Are tests for some traits more important than others? Why?
 - Would you want to know the results from a genetic test if you knew the contribution to the disease was about 50% genetic and 50% environmental? Why or why not?
 - How might bioethics play a role in this scenario?
 - How might bioinformatics play a role in this scenario?
 - What are some harms that could come from genetic testing?
 - What are some benefits that could come from genetic testing?
 - How is this skit realistic?
 - How is this skit not realistic?
 - Do you think that gene machines currently exist?

Possible answers can be found on Teacher Resource—*Discussion Questions for Meet the Gene Machine*.

Day 2

PART II: Direct-to-Consumer Genetic Testing

12. Ask students, “Do you think genetic tests are currently available for each of the traits mentioned in the *Meet the Gene Machine* play?”
13. Go through the list of genetic characteristics one by one and let students vote informally (thumbs up/thumbs down) as to whether they think genetic tests are currently available for each genetic trait mentioned in the play. The characteristics are:
 - Hair color
 - Musical ability

- Red blood cell count
- Height
- Carrier status for cystic fibrosis
- Predisposition towards breast cancer
- Predisposition towards alcoholism

14. Then, ask students to vote (thumbs up/thumbs down) as to whether they think information about the following additional traits is available through genetic testing:

- Sickle-cell anemia
- Earwax type
- Prostate cancer susceptibility
- Eye color
- Restless leg syndrome
- Mitral valve disease in dogs
- Resistance to HIV/AIDS

15. Tell students that they can find the answers to the above questions by visiting a number of websites. Today, the class will explore the website 23andMe, which offers **direct-to-consumer genetic testing**.

16. Using Teacher Resource—*Direct-to-Consumer Genetic Testing Website Exploration* as a guide, project the website for 23andMe (<http://www.23andme.com>) and explore it together as a class.

17. Teacher Resource—*Direct-to-Consumer Genetic Testing Website Exploration* has four parts:

- **Part A** exposes students to the variety of genetic tests available directly to the consumer.
- **Part B** distinguishes between Clinical Reports and Research Reports and asks if each test has equal **clinical validity** (these terms are defined in the Teacher Resource).
- **Part C** introduces the Breast Cancer Susceptibility Genes (BRCA) to students as **tumor suppressor genes** that normally prevent cancer. Mutations in these genes can lead to hereditary breast cancer and ovarian cancer when the normal functions of the proteins the genes encode are lost.
- **Part D** is recommended as homework if student have access to the internet at home. This section gives students background on how genetic testing works and contains a number of helpful animations.

PART III: Video Segment—A Family Disease

18. After exploring 23andMe as a class, remind students that genetic conditions touch the lives of people every day, including students in this classroom. Toward the end the lesson, ground students in a family's story about the repercussions of an inherited genetic disease and the options for genetic testing by sharing a video segment.

19. Show the eight-minute video segment "A Family Disease," Scene #14, from the NOVA video *Cracking the Code of Life*.

Direct-to-consumer genetic tests:

Direct-to-consumer genetic testing means an individual can receive information about his or her genetic condition without the use (or support) of a doctor or genetic counselor by submitting a DNA sample directly to the genetic testing company.

Clinical validity: How accurately a test predicts whether or not a person will get a particular disease or symptom (known as the "clinical outcome").

Tumor suppressor genes: Genes that encode proteins that help protect the cell from one step on the path to cancer. When both copies of this gene are mutated to cause a loss or reduction to their function, the cell can progress to cancer.


BRCA1: BReast **CA**ncer Susceptibility Gene **1**. *BRCA1* codes for the BRCA1 protein, which helps repair DNA damage and functions as a tumor suppressor.

20. After the video, tell students that they will be applying what they are learning about bioinformatics and genetic testing to a specific disease during the rest of the unit: inheritable breast cancer caused by mutations in one of the Breast Cancer Susceptibility genes (BRCA genes), specifically **BRCA1**.

Closure: *Careers in the Spotlight*

21. Close the lesson by returning to the picture of the bioengineer from the Lesson One PowerPoint Slides (**Slide #1**).

Bioinformatics & Genetic Testing: **Slide #1**



BIOENGINEER

ADRIENNE R. MINERICK, PhD

Place of Employment:
Michigan Technological University

Type of Research:
Biomedical Microdevices
For example, tiny devices to measure and manipulate living cells

"If a person is determined to learn, there will always be opportunities or resources for that person to pursue an education in science and engineering... There is a *real* demand for scientists and engineers whose contributions advance knowledge, technology, and the economic foundation of our society. I chose my career because I wanted to be a part of advancing knowledge and facilitating others to gain knowledge."

22. Show **Slide #2**, which provides job information for a bioengineer. Review this information with students.

Bioinformatics & Genetic Testing: **Slide #2**

CAREERS IN SPOTLIGHT: BIOENGINEER

What do they do?
A bioengineer uses the principles and tools of engineering to address problems in biology and medicine, creating usable products. These include designing medical devices, diagnostic equipment, renewable bioenergy, and genetically modified organisms.

What kind of training is involved?
A bachelor's degree in engineering is required for almost all entry-level engineering jobs. Sometimes a graduate degree is also required, such as a Master's degree or PhD

What is a typical salary for a Research Scientist?
The average salary is about \$55,000/year (\$26/hour), with a range from \$45,000/year (\$22/hour) to more than \$120,000/year (\$58/hour).

23. Tell students that today they were introduced to genetic testing and bioinformatics through the play *Meet the Gene Machine*, and by exploring the direct-to-consumer genetic testing company 23andMe. Bioengineers like Dr. Minerick are instrumental to gene testing in many ways, including:

- Much of our understanding about the functions of genes and their associations with diseases is made possible by laboratory equipment designed by bioengineers. This includes machines to help us purify sequences and analyze DNA, which are both necessary steps when performing genetic tests.

- If we someday have a “Gene Machine,” it will likely be a bioengineer who designs it and makes it!
24. Genetic testing today requires much more time to purify and analyze the patient’s DNA than shown in *Meet the Gene Machine*. In addition, one cannot simply look at a stretch of A’s, T’s, C’s, and G’s and know that it codes for a specific trait. This is why scientists use bioinformatics to help make sense of this vast amount of data.
 25. Ask students to answer Bioengineer Question #2 on their *Careers in the Spotlight* handout, which has students explain how this lesson has changed their understanding about the kind of work a bioengineer does.
 26. Ask students to also answer Bioengineer Question #3 on their *Careers in the Spotlight* handout, which has students explain how a bioengineer might use bioinformatics in his or her work.
 27. Tell students to keep their *Careers in the Spotlight* handout available for future lessons.

Homework

The following are suggested homework activities to follow this lesson:

- A. Ask students to write about the activities they learned in *Lesson One* in their lab notebooks, on another sheet of paper, or in a word processing program like Notepad or Microsoft Word® which they then provide to the teacher as a printout or via email. This can serve as an entry ticket for the following class. Have them complete these prompts:
 - a. Today I learned that...
 - b. An important idea to think about is...
 - c. Something that I don’t completely understand yet is...
 - d. Something that I’m really confident that I understand is...
- B. For students with internet access at home, Part D of Teacher Resource—*Direct-to-Consumer Genetic Testing Website Exploration* can be given as homework. Direct students to explore the “How it Works” section of 23andMe (<http://www.23andMe.com>). Students should click on “Genetics 101” and watch and take notes on all four short animations found in the “Genetics” section. Students can be encouraged to explore other interesting videos and animations on this page as well.
- C. To allow students additional time to explore the 23andMe website, an optional homework assignment, Student Handout—*Direct-to-Consumer Genetic Testing Homework: 23andMe*, may be assigned to be completed in-class or outside class, depending upon computer access and time available.
- D. For students without internet access at home, students can write a paragraph reflecting on the family shown in the video. If the student were in the daughter’s position, what would he or she do? Why?

[**Note:** Suggested scoring for homework: +5 points if all four prompts are complete.]

Glossary

Bioinformatics: Bioinformatics is the application of computer science and information technology to biology and medicine. Bioinformatics makes it possible to analyze large amounts of complex biological data and can be used to search biological databases, compare sequences, and draw molecular structures. Bioinformatic techniques are used to design and carry out the computer-based portion of genetic tests.

BRCA1: BReast **CA**ncer Susceptibility Gene **1**. *BRCA1* codes for the BRCA1 protein, which helps repair DNA damage and functions as a tumor suppressor.

Clinical validity: How accurately a test predicts whether or not a person will get a particular disease or symptom (known as the “clinical outcome”).

Direct-to-consumer genetic tests: Direct-to-consumer genetic testing means an individual can receive information about his or her genetic condition without the use (or support) of a doctor or genetic counselor by submitting a DNA sample directly to the genetic testing company.

Established research report: Established Research Reports from 23andMe provide information about conditions and traits for which there are genetic associations supported by multiple, large, peer-reviewed studies. Because these associations are widely regarded as reliable, 23andMe uses them to develop quantitative estimates and explanations of what they mean for individuals receiving direct-to-consumer genetic tests.

Genetic testing: The analysis of a person’s DNA. It is usually done to determine whether that individual carries changes (mutations) to genes that make them more susceptible to a disease. It is also popularly used to find out about ancestry and paternity.

Preliminary research reports: Preliminary Research Reports from 23andMe are based on peer-reviewed, published research for which the findings still need to be confirmed by the scientific community. They also include topics where there may be contradictory evidence. Topics may move from Preliminary Research to Established Research when and if sufficient follow-up studies are performed.

Tumor suppressor gene: Gene that encodes a protein that helps protect the cell from one step on the path to cancer. When both copies of this gene are mutated to cause a loss or reduction to their function, the cell can progress to cancer.

Resources

For more information about polymerase chain reaction, DNA sequencing, and microarray technologies, the Howard Hughes Medical Institute (HHMI) “Bioactive” website has a large collection of useful videos and animations on a number of topics. These can be freely accessed online at: <http://www.hhmi.org/biointeractive/video/index.html>.

Credit

Minerick, Adrienne Robyn. Personal Interview. 29 September 2010.

“Meet the Gene Machine” was written by Laura Streith, Karen Bultitude, Frank Burnet, and Claire Wilkinson at the Science Communication Unit at the University of the West of England and was funded by Wellcome Trust, United Kingdom. Adapted and used with permission.

The two-hour NOVA video *Cracking the Code of Life* can be purchased at Amazon.com or through the PBS website at: <http://www.shoppbs.org/product/index.jsp?productId=2980750>. Although many genetic advances have occurred since the video’s 2001 release date, many of the video segments are relevant to this curriculum.

The NCBI resource on genetic tests called “Gene Tests” can be found at: <http://www.ncbi.nlm.nih.gov/sites/GeneTests/>.

Name _____ Date _____ Period _____

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Careers in the Spotlight

What is bioinformatics? Bioinformatics is the application of computer science and information technology to biology and medicine.



<p>Lesson One Career: Bioengineer Adrienne R. Minerick, PhD</p> <p>1. What kind of work do you think a bioengineer does?</p>	<p>2. How has this lesson changed your understanding about the kind of work a bioengineer does?</p> <p>3. How do you think a bioengineer might use or benefit from bioinformatics?</p>
<p>Lesson Two Career: Veterinarian Deborah Tegarden, DVM</p> <p>1. What kind of work do you think a veterinarian does?</p>	<p>2. How has this lesson changed your understanding about the kind of work a veterinarian does?</p> <p>3. How do you think a veterinarian might use or benefit from bioinformatics?</p>
<p>Lesson Three Career: Genetic Counselor Robin Bennett, MS</p> <p>1. What kind of work do you think a genetic counselor does?</p>	<p>2. How has this lesson changed your understanding about the kind of work a genetic counselor does?</p> <p>3. How do you think a genetic counselor might use or benefit from bioinformatics?</p>

LESSON 1

HANDOUT



Lesson Four Career: Laboratory Technician

Zane Kraft, MS

1. What kind of work do you think a **laboratory technician** does?

2. How has this lesson changed your understanding about the kind of work a **laboratory technician** does?

3. How do you think a **laboratory technician** might use or benefit from bioinformatics?

Lesson Five Career: 3D Animator

Beth Anderson

1. What kind of work do you think a **3D animator** does?

2. How has this lesson changed your understanding about the kind of work a **3D animator** does?

3. How do you think a **3D animator** might use or benefit from bioinformatics?

Lesson Six Career: Bioethicist

Kelly Edwards, PhD

1. What kind of work do you think a **bioethicist** does?

2. How has this lesson changed your understanding about the kind of work a **bioethicist** does?

3. How do you think a **bioethicist** might use or benefit from bioinformatics?

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Meet the Gene Machine

*Modified from "Meet the Gene Machine," written by **Laura Strieth**, **Karen Bultitude**, **Frank Burnet**, and **Clare Wilkinson**. "Meet the Gene Machine" was devised by the Science Communication Unit at the University of the West of England, Bristol, and is funded by the Wellcome Trust (adapted with permission).*

A daytime tv talk show set. Sofa, coffee table, vase of flowers, gene machine (covered). Host and scientist are preparing to go live. Host is standing at the sofa, checking makeup and hair, which requires more concentration than talking to the scientist. Host is stressed and short-tempered at first, but becomes relaxed and happy when on air, brimming with confidence. Scientist is nervous, but excited. They are not yet on the air.

Host	Come on you guys; two minutes is all!! Are these flowers for real? Move that boom, it's right in my line of sight, and has anybody seen my next guest?!!
Chris	Hello, I am Dr. Chris Taylor, I'm a scientist from...
Host	Great, (INDICATING SOFA) you sit at that end. You know who I am of course. Have you done much TV?
Chris	No, this is my first time, actually I'm feeling a bit...
Host	Great, it's a piece of cake. You'll love it. Did you bring the gizmo?
Chris	Yes, it's here. Should I get it out?
Host	We have a saying in TV; don't tell me, show me. Better make it quick, we've got two minutes for the break then we're back with you as first item. What do you guys call that thing?
Chris	Oh, it's called the <i>Microarray and Microassay Hyper Channel Optimizing Genetic Analysis with Real Time Interpretive Functionality Unit</i> .
Host	OK... the Gene Machine and what does it do?
Chris	We'll take a sample of DNA, insert it in here, and the Microarray and...
Host	Gene Machine.
Chris	...and the Gene Machine analyzes it and prints out a complete genetic profile, which I'll help to interpret.
Host	Interpret? What's it speak, German?
Chris	No, but the results can be confusing. Very few things in genetics are black and white and most people need help figuring out what the test results mean to them.
Host	Do you think I got my own talk show by being "most people"? No, I got here by having the newest, most exciting gadgets on my show. Did you see Jerry has a lie detector on his show? Hah, we got genetic tests – how cool is that?

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Chris It's not a toy.

Host Is a lie detector a toy? This is reality, not a game show. One minute left – how does it work?

Chris It looks at the 25,000 genes that play a vital role in making us who we are; things like hair color, height, aspects of personality all have some genetic basis.

Host So if we've all got these genes that control everything, how do we end up so different?

Chris There are many, many different versions of genes out there and we all inherit different combinations from our parents. Only identical twins have the same genes.

Host Is that why they always dress the same?

Chris When they get the choice they don't. Identical twins may seem similar, but actually, there are lots of differences between them.

Host So how does that happen?

Chris Our genes are only part of the story. What happens to us in our lives also makes a difference to us.

Host Kind of "Inheritance vs. Circumstance," no, how about "Biology vs. Experience?"

Chris It's usually called "Nature vs. Nurture."

Host Hey, that's good – are you looking for a job?

Chris I have one.

Host And now we have the Gene Machine. This is great. So, where do you get the DNA?

Chris That's the easy part; I just take a sample with this cotton swab.

Host "The Cotton Swab of Destiny." Go for it.

Chris What?

Host Fifteen seconds, grab the DNA.

Chris From you?

Host Who else? This is my show.

Chris I need you to sign a consent form.

Host I don't sign anything without a lawyer.

Chris Are you sure about this?

Host Didn't I say so?

Chris Well...

Host Five seconds.

Scientist takes a swab of the inside of host's cheek.

Host Places everyone! (TO SCIENTIST) Do something with that hair.

Show prepares to go live. 3 ... 2 ... 1 ... We are live. Host turns casually from Chris delighted to see the audience back for more. Host is relaxed and confident. Chris is flustered and adjusting hair.

Host Welcome back; so good to see you all again. Now, stop what you're doing; sit down and pay attention, because we are about to make television history. Ladies and Gentlemen, please meet the Gene Machine.... (LEADS THE APPLAUSE)

Applause

Host And with me today is scientist, Dr. Chris Taylor.

Chris Hello.

Host Chris, we are all just so excited about having you and the Gene Machine here with us today.

Chris (STILL HOLDING THE COTTON SWAB) It's great to be here.

Host Now Chris and other scientists have invented a fantastic new device that can read all your genes.

Chris Yes, It's called the Microarray...

Host The Gene Machine, that's right Chris. Now as I understand it Chris, we all have genes inherited from our parents.

Chris Yes, we actually have two full sets of genes, one from each parent. One from the sperm and one from the egg.

Host OK, OK, keep it clean doctor, this is a family show. Now, these genes can tell us a lot of things, can't they?

Chris Yes, our genes determine many things about us, many of our key characteristics.

Host So the Gene Machine can tell us much more than, for instance, a lie detector?

Chris Oh yes, a lie detector will only tell you if somebody is lying.

Host Whereas we will know the truth.

Chris (SEES DISASTER LOOMING) The output from the machine is true, yes. But it can only tell us about a person's genes....

Host You bet. In fact, this will change the whole future of reality television. With this new machine we can get closer to the truth than ever before.

Chris Errm, it's not just for television.

Host (WORRIED) Oh? Who else has a Gene Machine?

Chris Well, medicine obviously.

Host Really?

Chris Yes, many common diseases have genetic components. Identifying those genes will mean doctors can treat people more effectively.

Host Isn't that great!

Chris Obviously the police and others would like to use the machine to assist in identifying people...

LESSON 1

HANDOUT

Host But the biggest impact will be here on TV. And guess who will be the very first celebrity to Meet the Gene Machine...

Chris You?

Host (LAUGHS) As we like to say in television "Here's one I prepared earlier." (INDICATES MACHINE)

Chris Oh yes, we did. I have it here.

Operates machine. The swab goes in one side and a large printout comes out the other side.

Host Wow, that's a lot of paper. More is always better, right?

Chris (TRYING TO HOLD THE PRINTOUT) Well, we all have the same amount of genetic information, but there is a lot of it. Even if genetic tests only look a small part of each gene, that is still quite a bit of information.

Host So what does it say?

Chris Well first of all, it confirms that that is your natural hair color, and will be until you are in your 50s. However long that may be.

Host Does it tell you my age?

Chris No, your genes stay more or less the same all through your life.

Host So I have the genes of a teenager? (CHUCKLE) What else can you see there?

Chris (JUGGLING THE PAPERS) Ah, you have a number of gene variants associated with being musical.

Host I knew it!! I always said I had talent. My mom said I was too lazy to have lessons.

Chris You have some versions of some genes that are associated with musical ability. But, yes, hard work and practice are still very important.

Host I could have been a superstar – wait a minute, I AM.

Chris (TRIES TO ENJOY THE JOKE)

Host OK, so what else have you got there?

Chris Well, it says here that you are likely to be quite tall.

Host (BECOMING IMPATIENT) I can see that for myself; let me have a look. (TAKES THE PRINTOUT)

Host What's this?

Chris That says that you produce a lot of red blood cells.

Host So?

Chris So you should have high levels of fitness and stamina.

Host OK. Tell me something I don't know. What's this?

Chris Oh, that's quite common.

Host So what does it do?

Chris We can talk about that after the show.

Host I'm happy to share this with my public.

Chris That says you carry the gene variant that causes cystic fibrosis.

Host Now you're wrong there, I'm as fit as a fiddle, you just said so.

Chris No, it's not like that. I said earlier that genes come in different varieties, and some varieties cause harm. If you had inherited this variety from each parent, then you would have the disease.

Host So I'm OK?

Chris Yes, but you could pass the gene on to your own children, and the disease too, if your partner also has this version of the gene.

Host How would I know?

Chris Ask your partner to use the machine.

Host Oh boy, our lawyers are gonna love you. OK, what does this one mean?

Chris Ah, now that's a really interesting gene.

Host Uh huh.

Chris Well, it's pretty technical.

Host You don't think my audience is stupid, do you?

Chris No, no, no, it's just...

Host Yes?

Chris (DEEP BREATH) That gene suggests you have a predisposition towards breast cancer.

Host What's a predisposition?

Chris Well, it doesn't mean you will get the disease, but you might.

Host Anybody might.

Chris Yes, but you are at greater risk.

Host How much greater?

Chris That would be hard to say.

Host So what's the point in telling me? It's in my genes, so there's nothing I can do

Chris There are lots of things you can do to reduce the risk: diet, exercise, self examination. How often do you examine your breasts?

Host (NO LONGER HAVING FUN) You scientists really are quite blunt aren't you? We've got time for one more result before the break. How about this one?

Chris That one?

Host Yeah.

Chris (INDICATES ANOTHER) This one's interesting.

Host That one.

Chris It's sort of private.

LESSON 1

HANDOUT

Host I have nothing to hide.

Chris Are you sure?

Host It's a disclosure show – get disclosing.

Chris You are at very high risk of alcohol dependence.

Host What is that supposed to mean?

Chris Alcohol dependence is a condition in which a person...

Host (STANDS UP) I know what an alcoholic is.

Chris (STANDING UP) Look, you asked me to tell you.

Host Are you trying to ruin my reputation?

Chris Of course not, in fact I tried to warn you about revealing this information in public.

Host You didn't tell me it would be like this. This is terrible.

Chris The information in our genes is very powerful, and valuable.

Host Yes, to blackmailers and paparazzi.

Chris No, to all of us.

Host You want to do this to everyone?

Chris Of course not, but we can't keep this information locked away forever. Gene testing is here to stay.

Host People have a right to privacy!

Chris People have a right to know!

Host To know how much? To know about who? Who decides?

Chris The American public of course, but do you have the nerve to ask them what they think?

Host Yes, I do.

1

Direct-to-Consumer Genetic Testing Homework: 23andMe

Aim: To understand what type of genetic information a for-profit company is selling to the general public and determine the accuracy of that information.



Instructions: Go to the complete list of health reports at the 23andMe genetic testing website at <https://www.23andme.com/health/all/>. Answer these questions on a separate sheet of paper and turn in your answers to your teacher.

1. In your own words, describe an Established Research Report.
2. Find a condition that has an Established Research Report (there is a * after the name). Write that condition's name on your paper, and click the link to the report for that condition.
3. Using the information under the "Example Data" tab, in your own words, summarize the description paragraph of the disease or condition.
4. Look at the "Genes vs. Environment" section of the report – what does it say about the heritability of your condition and how much genetics might play a part in it?
5. Often, the Example Genetic Data is given in terms of % men of European ethnicity. However, the condition may actually affect other populations more. Read the **Description** and **Genes vs. Environment** sections to find if it is currently known that your condition affects certain populations more than others. If so, explain which populations.
6. Go back to the "Health Reports: Complete List." In your own words, describe a Preliminary Research Report.
7. Find a condition that has a Preliminary Research Report. Write down the name of that condition and click the link to the report for that condition.
8. Using the information under the "Example Data" tab, in your own words, summarize the description paragraph about the disease or condition and describe how much genes contribute to the condition.
9. Are Established Research Reports or Preliminary Research Reports more scientifically reliable? Explain.
10. Clinical validity is a term that bioethicists, doctors, and genetic counselors use to describe how accurately a genetic test predicts whether a person will get a particular disease or symptom. Are all the tests offered on 23andMe equally clinically valid? Explain.

1 Understanding Genetics and SNPs

Aim: To use educational animations developed by 23andMe to learn more about genetics and direct-to-consumer genetic testing.



Instructions: Go to the 23andMe website <https://www.23andme.com/>. Follow the instructions below and answer the questions on a separate sheet of paper. Turn your answers in to your teacher.

1. Click the "How it Works" button under the green bar at the top of the page.
 - a. How is this like the Gene Machine?
 - b. How is it different from the Gene Machine?
2. From the "How it Works" section, scroll down to the cartoon picture on the left, under "Genetics 101." Click to begin where it says "Watch an animated guide to your genes, SNPs, phenotype, and more."
 - a. Approximately how many genes does each human cell contain?
 - b. Approximately how similar are human beings to chimpanzees?
 - c. Approximately how similar are human beings to one another?
3. From the column on the left, under "Genetics," click "What are SNPs?" and "What is phenotype?" (See **Figure 1**).



Figure 1: Watch an animated guide. Credit: 23andMe, <https://23andme.com/>.

- a. What is a SNP? Explain not only what the abbreviation stands for, but also what a SNP is.
- b. Most genetic tests offered by 23andMe evaluate SNPs. Do all SNPs result in a change in phenotype? Explain.

1

Discussion Questions for Meet the Gene Machine

1. What genetic traits are mentioned in the play? (List these traits on the board for all to see.)

- Hair color
- Musical ability
- Red blood cell count (leading to high levels of fitness and stamina)
- Height
- Carrier status for cystic fibrosis
- Predisposition towards breast cancer and alcoholism

2. Are genetic tests for all of the traits mentioned in the play equally valuable? Are tests for some traits more important than others? Why?

Example: A genetic test to determine hair color is not very useful if hair color can be changed and the results do not have an effect on one's health. A test to determine whether or not a person carries the gene variant that causes cystic fibrosis can be important, especially if the disease is known to run in a partner's family.

3. The environment plays a large part in diseases, even genetic diseases. In the play, Chris and the host have this interaction:

Chris (DEEP BREATH) That gene suggests you have a predisposition towards breast cancer.
 Host What's a predisposition?
 Chris Well, it doesn't mean you will get the disease, but you might.
 Host Anybody might.
 Chris Yes, but you are at greater risk.
 Host How much greater?
 Chris That would be hard to say.
 Host So what's the point in telling me? It's in my genes, so there's nothing I can do.
 Chris There are lots of things you can do to reduce the risk: diet, exercise, self examination.

Would you want to know the results from a genetic test if you knew the contribution to the disease was about 50% genetic and 50% environment? Why or why not?

Answers will vary. Some students may choose not to know because they have no control over part of it, and some may choose to know because they do have control over part of it. Some may say it's worth reducing risk through a healthy diet and exercise even if they don't know the results from the genetic test.

4. How might bioethics play a role in this scenario?

Bioethics, and the use of bioethical principles (introduced to students in *Lesson Three*) can help students analyze difficult or conflicting issues in a systematic, rational way. Issues include the right to privacy versus the right for individuals and families to know about their genetic make-up.

5. How might bioinformatics play a role in this scenario?

The Gene Machine itself uses tools of bioinformatics. Genetic testing is possible through the use of bioinformatic tools. Students will use these tools in *Lessons Two, Four and Five*.

6. What are some harms that could come from genetic testing?

Answers may vary. For example, a person's genetic information might not remain private, as in the skit. A person might also be anxious, and not live life to the fullest, knowing that he or she has a predisposition to a genetic disease. There can also be harms associated with testing for a disease for which there is no cure.

7. What are some benefits that could come from genetic testing?

Answers may vary. For example, a person who has a predisposition to a disease can make positive health and environmental changes. People considering parenthood can screen for genetic diseases if a serious disease is known to run in the family.

8. How is this skit realistic?

Answers may vary. Many talk shows today show genetic testing, such as for paternity. However, there are challenges in translating knowledge from scientists to the public, and there are risks of over-simplifying genetic diseases. Like the host, many people don't realize how serious the information obtained from a genetic test may be. There are a number of known genetic diseases for which tests are available. According to the NCBI resource *GeneTests*, there are more than 1,500 genetic tests currently available, some of which students will learn more about by exploring 23andMe.

9. How is this skit not realistic?

Answers may vary. It is commonly assumed that scientists and doctors fully understand the functions of all 25,000 genes in the human genome. However, much research still needs to be done. In addition, many characteristics or phenotypes are the result of complex interactions between genes and environment, or the result of the interaction of many genes (called "multifactorial traits"), meaning simple genetic tests would be misleading. The test results were also provided much too quickly. Current sequencing technology would require days to produce this much data. Also, a signed consent form would be required before performing any genetic test. This form would provide information about the risks and benefits of the genetic test.

10. Do you think that gene machines currently exist?

Students may be familiar with biotechnology applications, including polymerase chain reaction and DNA sequencing. Some students may have seen the movie "GATTACA," in which a character provides a hair sample and receives a complete printout of her genome in a matter of minutes. However, there is currently no 'gene machine' like the one in this play. Bioengineers have helped develop many tools and machines that make genetic testing, as well as genetic research, possible. Genetic testing is currently a multi-step process: DNA is extracted using a machine like a centrifuge, the DNA is copied using a thermocycler and polymerase chain reaction (PCR), and then the DNA is sequenced using a DNA sequencing machine. Multiple genetic tests can be performed at the same time on the same sample using a process called DNA microarray, which can be used to detect single nucleotide changes in a given region of DNA. However, all of these types of genetic tests require much more time to perform than the genetic tests in this play. In addition, one cannot simply look at a stretch of A's, T's, C's, and G's and know that it codes for a specific trait. This is why scientists use bioinformatics to help make sense of this vast amount of data.

1

Direct-to-Consumer Genetic Testing Website Exploration

PART I: What Types of Genetic Tests are Available?

1. Go to the 23andMe homepage: <https://www.23andme.com/>. Ask students: **What are the types of DNA tests that 23andMe offers?**

Ancestry Edition and Health Edition.

2. Take a few minutes to become familiar with the site. Ask students: **What do you think 23andMe does? Who is it for?**

23andMe's mission is, "To be the world's trusted source of personal genetic information." They allow individuals to receive genetic information without the use of a doctor or medical professional. Their services are available to anybody who can pay the fee for service.

3. Return to the Welcome page, found on the upper left green bar. Scroll down to find and click the blue link to "see all 201 topics" to take you to the complete list of "Health Reports" (see **Figure 1**). 23andMe is adding new topics all the time, so this number may have increased by the time you view this website.

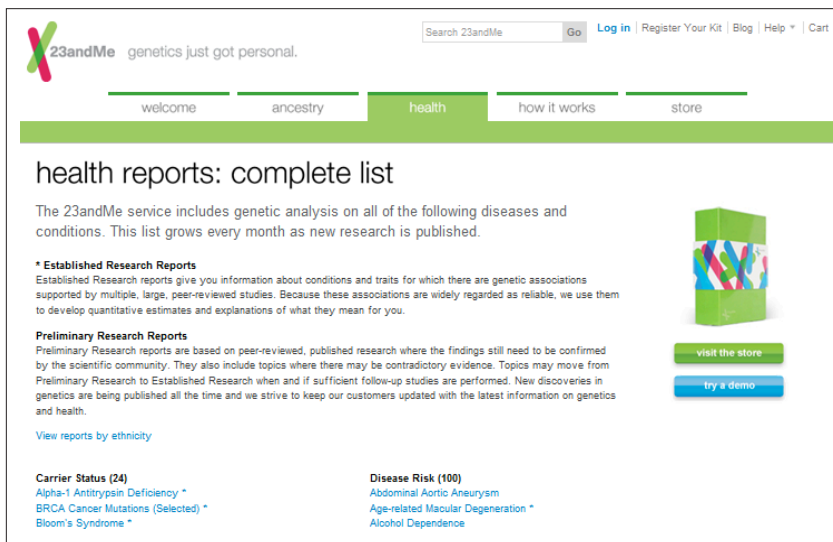


Figure 1: Health Reports.
Credit: 23andMe, <https://www.23andme.com/>.

4. One at a time, scroll through the list of Health Reports to find some of the genetic traits from the list that the class “voted” on earlier, such as Height (under “Traits”) or Sickle-Cell Anemia (under “Carrier Status”).

Does 23andMe have a genetic test for all of those traits?

23andMe tests for all of the diseases and traits mentioned in “Meet the Gene Machine,” except for musical ability, red blood cell count, and mitral valve disease in dogs. Information about mitral valve disease is available to veterinarians, using the tools of bioinformatics.

5. Look at the complete list of reports. **Which traits do students find most interesting?**

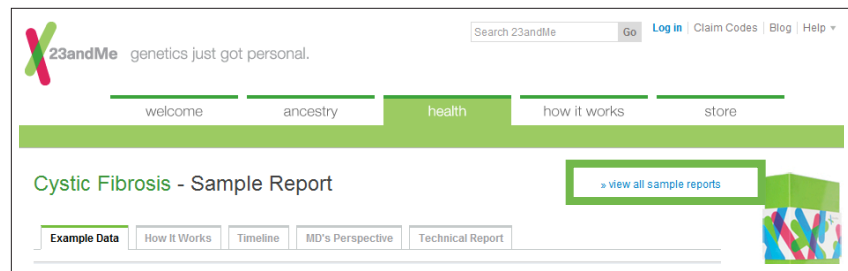
Answers will vary depending on students’ interests.

6. Click on some traits or diseases that are interesting to students, and/or that you have studied in class. Explore the tabs at the top of the page (Example Data, How it Works, Technical Report) and the video(s) or picture(s) on the page.

PART II: Established versus Preliminary Research Reports

7. From any sample report page, click on “>>view all sample reports” on the top right of the page to return to the complete list of health and trait topics (see **Figure 2**).

Figure 2: View all sample reports.
Credit: 23andMe, <https://www.23andme.com/>.



8. For any given Carrier Status, Disease Risk, Drug Response, or Trait, there are **Established Research Reports** and **Preliminary Research Reports**. Show students examples of each of these reports in the context of diseases or traits. Those with Established Research Reports are noted with asterisks*.

What is an Established Research Report? Example: Cystic fibrosis

According to 23andMe: “Established Research Reports give you information about conditions and traits for which there are genetic associations supported by multiple, large, peer-reviewed studies. Because these associations are widely regarded as reliable, we use them to develop quantitative estimates and explanations of what they mean for you.” In other words, there is good scientific evidence that the genes described in Established Research Reports are truly associated with a disease or condition.

What is a Preliminary Research Report? Example: Alcohol dependence

According to 23andMe: “Preliminary Research reports are based on peer-reviewed, published research for which the findings still need to be confirmed by the scientific community. They also include topics where there may be contradictory evidence. Topics may move from Preliminary Research to Established Research when and if sufficient follow-up studies are performed.” In other words, the associations described in Preliminary Research Reports represent initial findings and should be interpreted with caution.

Preliminary Research Reports:

Preliminary Research Reports from 23andMe are based on peer-reviewed, published research for which the findings still need to be confirmed by the scientific community. They also include topics where there may be contradictory evidence. Topics may move from Preliminary Research to Established Research when and if sufficient follow-up studies are performed.

Which type of report is supported with more reliable science?

Established Research Reports. Emphasize the importance of higher **clinical validity**, and information gained from large, peer-reviewed studies which can be supported by more than one study. A test with high clinical validity means that the test is very good at predicting whether someone will get the disease or condition.

Are all the genetic tests offered at 23andMe equally clinically valid?

No. The scientific community has not yet agreed about the level of meaningfulness of these tests. The tests can vary greatly in their ability to predict disease. Because of this variability, results of these tests can be difficult to interpret.

PART III: Breast Cancer Susceptibility Genes

9. Give students some background about the Breast Cancer Susceptibility (BRCA) genes. Tell students that this unit of study focuses on one gene—**BRCA1**—that plays a crucial role in DNA repair. *BRCA1* is a tumor suppressor gene that normally prevents cancer. Mutations in this gene can lead to hereditary breast cancer and ovarian cancer when the normal function is lost. BRCA is sometimes pronounced “BRACK-uh.”
10. Tell students that there are two BRCA genes, *BRCA1* and *BRCA2*. This unit focuses primarily on *BRCA1*.
11. Click on “BRCA Mutations (Selected)” from the complete list of health reports from above (Part B). Read the note in the peach-colored box (see **Figure 3**). ***How complete is the BRCA test at 23andMe?***

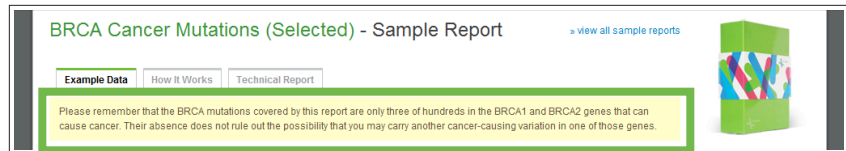
23andMe tests for only three of the hundreds of known mutations in the *BRCA1* and *BRCA2* genes. These *BRCA1* and *BRCA2* genes have been patented by the biopharmaceutical company Myriad Genetics Incorporated. Myriad has also patented the method of testing the genes for mutations, so much of the *BRCA1* and *BRCA2* information is unavailable to 23andMe.

BRCA1: BReast **C**ancer Susceptibility Gene 1. *BRCA1* codes for the BRCA1 protein, which helps repair DNA damage and functions as a tumor suppressor.

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Figure 3: Read the note in the peach box.
Credit: 23andMe, <https://www.23andme.com/>.



If you suspected mutations to the BRCA1 and BRCA2 genes ran in your family, would you choose to get tested at 23andMe or through Myriad?

Myriad is considered the gold standard for *BRCA1* and *BRCA2* testing since they have a patent on the gene and therefore have unhindered access to the genetic information.

12. Continue scrolling through the BRCA Cancer Mutations Sample Report. As a class, read the “About Breast/Ovarian Cancer” section.

Direct-to-Consumer Genetic Testing (continued)

[**Note:** Omit this question if students have not learned about types of mutations.]

13. Scroll down to the “Example Genetic Data.” ***Drawing on previous knowledge, what are the most common types of mutations?***

Insertions, substitutions, and deletions.

What type of mutation might 185delAG be?

The del refers to a deletion mutation. (See **Figure 4.**)

What else can you tell from the name of the mutation?

At nucleotide #185 in the DNA sequence, there is a deletion of an A and a G—an Adenosine and a Guanine.

Figure 4: The *del* refers to a deletion mutation.
Credit: 23andMe, <https://www.23andme.com/>.

Example Genetic Data	
Who	What It Means
	Carrier for the 185delAG BRCA1 mutation. Lifetime risk of breast cancer for women is increased from 13% to 81% and risk of ovarian cancer is increased from less than 2% to 54%. May significantly increase risk of prostate cancer in men. There is also an increased risk for breast cancer in men.

14. Scroll down to look at the other two mutations on the “Example Genetic Data.” **What does 5382insC mean?**

At nucleotide #5382, a Cytosine is inserted.

What does 6174delT mean?

At nucleotide #6174, a Thymine is deleted.

15. Scroll down the page to find the section on “Citations.” **Do these citations look to be from valid, scientific sources?**

Yes. *Science* magazine, *New England Journal of Medicine*, and *American Journal of Human Genetics* are all well-respected, peer-reviewed sources.

16. Point out that the research cited is done with equipment designed by **bioengineers**, our career of interest for the day.

PART IV: How it Works

[Optional: Could Be Assigned for Homework Using the Student Handout—*Understanding Genetics and SNPs*]

17. Click the “How it works” button under the green bar at the top of the page. **How is this like the Gene Machine?**

A genetic sample (saliva) is sent to 23andMe for analysis and results are given to the client.

How is it different from the Gene Machine?

It takes two to four weeks to analyze the sample; the results are not instant, as with the Gene Machine. Although not shown, the process requires a number of steps, not one machine. 23andMe looks at over 200 traits and topics; it is unclear what is (or is not) analyzed with the Gene Machine.

18. Scroll down to the cartoon picture on the left, under “Genetics 101.” Click to begin where it says “Watch an animated guide to your genes, SNPs, phenotype, and more.”

19. From the column on the left, under “Genetics,” click any of the four animation titles. Of specific interest are “What are SNPs?” and “What is phenotype?” (See **Figure 5**.)



Figure 5: Click on the four animation titles.
Credit: 23andMe, <https://www.23andme.com/>.

LESSON 1

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1

Answers for Direct-to-Consumer Genetic Testing Homework: 23andMe

[**Note:** Suggested point values are included after each question, and are intended to provide general guidelines for the weight each question could be given. Using these suggested point values, the total value for this worksheet is **10 points**.]

1. In your own words, describe an Established Research Report.

An Established Research Report contains information from **multiple, large peer-reviewed scientific studies**. A peer-reviewed study has been examined by the scientific community and the research methods and conclusions of the study authors has been found to be appropriate. These reports contain more reliable information than "Preliminary Reports."

(+1 for including a description or for explaining that they are more reliable than preliminary reports.)

2. Find a condition that has an Established Research Report (there is a * after the name). Write that condition's name on your paper, and click the link to the report for that condition.

Student answers will vary.

3. Using the information under the "Example Data" tab, in your own words, summarize the description paragraph of the disease or condition.

Student answers will vary based on their answer to Question #2.

(+1 for an accurate description of the condition.)

4. Look at the "Genes vs. Environment" section of the report – what does it say about the heritability of your condition and how much genetics might play a part in it?

Student answers will vary based on their answer to Question #2.

However, many conditions have both genetic and environmental factors.

(+1 for statement about the heritability of the condition; +1 for further explanation of genetic contribution.)

5. Often, the Example Genetic Data is given in terms of % men of European ethnicity. However, the condition may actually affect other populations more. Read the **Description** and **Genes vs. Environment** sections to find if it is currently known that your condition affects certain populations more than others. If so, explain which populations.

Student answers will vary based on their answer to Question #2 above. However, several conditions are more prevalent in certain populations.

(+1 for listing populations more affected or for answering 'No, it is not currently known.')

6. Go back to the “Health Reports: Complete List.” In your own words, describe a Preliminary Research Report.

A Preliminary Research Report contains information from one or a few scientific reports that have been peer-reviewed (reviewed by the scientific community), but there is not sufficient data to confirm the results of the study. There may be cases in which two studies about the same gene(s) or condition(s) are actually contradictory. Once enough data and studies have been conducted to confirm an association, Preliminary Research Reports may become Established Research Reports.

(+1 for including bolded ideas or indicating that information comes from a small number of studies.)

7. Find a condition that has a Preliminary Research Report. Write down the name of that condition and click the link to the report for that condition.

Student answers will vary.

8. Using the information under the “Example Data” tab, in your own words, summarize the description paragraph about the disease or condition and describe how much genes contribute to the condition.

Student answers will vary based on their answer to Question #6. However, many conditions have both genetic and environmental factors.

(+1 for accurate description of condition; +1 for description of relative contributions of genes and environment.)

9. Are Established Research Reports or Preliminary Research Reports more scientifically reliable? Explain.

Established Research Reports are more reliable than Preliminary Research Reports, because they draw on information from multiple, large, peer-reviewed scientific studies. Preliminary Research Reports are based on one, or only a few, scientific studies which often contain fewer research subjects.

(+1 for Established Research Reports, with explanation.)

10. Clinical validity is a term that bioethicists, doctors and genetic counselors use to describe how accurately a genetic test predicts whether a person will get a particular disease or symptom. Are all the tests offered on 23andMe equally clinically valid? Explain.

No, all of the tests offered on 23andMe are not equally clinically valid. For example, conditions for which there are Established Research Reports tend to be more clinically valid than conditions for which there are only Preliminary Research Reports. In addition, some students may note that some genetic associations only account for a small portion of the risk of a disease or condition (traits for which heritability is low). In this case, if a particular gene or allele only conveys a small portion of the risk, the genetic test for that condition may not be considered clinically valid. Students will explore this issue in depth in Lesson Six.

(+1 for reference to Established Research Reports; +1 for reference to different genetic contributions.)

1

Understanding Genetics and SNPs

[Note: Suggested point values are included after each question, and are intended to provide general guidelines for the weight each question could be given. Using these suggested point values, the total value for this worksheet is **7 points**.]

1. Click the "How it Works" button under the green bar at the top of the page.

a. How is this like the Gene Machine?

A genetic sample (saliva) is sent to 23andMe for analysis, and results are given to the client. (+1 point.)

b. How is it different from the Gene Machine?

It takes two to four weeks to analyze the sample; the results are not instant, as with the Gene Machine. Although not shown, the process requires a numbers of steps, not one machine. 23andMe looks at over 120 traits and topics; it is unclear what is (or is not) analyzed with the Gene Machine. (+0.5 for each difference noted between the Gene Machine and 23andMe, up to +1.5 points.)

2. From the "How it Works" section, scroll down to the cartoon picture on the left, under "Genetics 101." Click to begin where it says "Watch an animated guide to your genes, SNPs, phenotype, and more."

a. Approximately how many genes does each human cell contain?

20,000. (+0.5 point.)

b. Approximately how similar are human beings to chimpanzees?

Human beings and chimpanzees share 98.5% of their genes. (+0.5 point.)

c. Approximately how similar are human beings to one another?

Human beings share 99.5% of their genes with one another. (+0.5 point.)

3. From the column on the left, under "Genetics," click "What are SNPs?" and "What is phenotype?"

a. What is an SNP? Explain not only what the abbreviation stands for, but also what an SNP is.

A SNP is a single nucleotide polymorphism. A SNP is a single base pair change among individuals. (+1 for explaining what "SNP" stands for; +1 for the explanation of what an SNP is.)

b. Most genetic tests offered by 23andMe evaluate SNPs. Do all SNPs result in a change in phenotype? Explain.

Most SNPs do not result in a change in phenotype. These are "silent" changes that do not result in a different amino acid within the coding sequence of a protein. (+0.5 points for noting that all (or most) SNPs do not result in a change in phenotype; +0.5 points for explaining why.)

LESSON 1

KEY