SNAPSHOT

Direct-To-Consumer Testing in the Genomic Age

Teacher Guide

Adapted for PBS LearningMedia in partnership with WETA for use with

KEN BURNS PRESENTS
THE GENE
AN INTIMATE HISTORY

Big Picture:

The effort to read the human genetic code – to identify and sequence the ~20,000 human genes (and all the DNA in between) – was a huge accomplishment. Known as the Human Genome Project, it has laid the foundation for many new discoveries, tools, and medical tests that have emerged in the years since its completion in 2003.

Technological developments are making it possible to read a person's entire genetic code, or genome, more rapidly and at a lower cost than ever before. Personal genome sequencing is allowing scientists and doctors to better understand the connections between genes and human health, improve medical care, help extend people's lives, and provide information about our shared human ancestry. As the cost of genetic analysis decreases and research advances, it is increasingly possible to include a person's genetic make-up in the toolkit that informs their healthcare.

The growing field of personal genetics is at the intersection of science and society. It is an exploration into the complex interactions through which our genes and our environment influence our physical, mental and behavioral states. As this field moves forward, conversation is needed about the meaning for individuals and society.

Created by the Personal Genetics Education Project (pgEd.org).
Adapted for PBS LearningMedia in partnership with WETA (2020) – bit.ly/GeneEducation

Foundational concept

An important theme throughout this lesson is that your physical, mental and behavioral traits are influenced both by your genetic make-up and by the unique circumstances in which you have lived your life, including everything that has ever happened to you. We often refer to these two inputs as "nature," the genome you carry that is unique to you, and "nurture," the environment in which you have lived your life. However, "nature" and "nurture" are not independent of each other – environmental factors affect how genes are "expressed" (turned on or off), and even lead directly to changes in your DNA sequence. The intricate and sometimes unclear relationship between genetics and environment is key to our understanding of behavior and health. Researchers are seeking to study the genome sequences and characteristics of many people to develop a better understanding of the interplay of nature and nurture in known diseases, with the hope that this would lead to better treatments, cures, preventative measures and healthier generations of children.

Activities:

Part 1: Do Now Exercise (7 minutes)

Have students answer the three questions listed on slide 2 in the slideshow, either individually or in pairs. These questions are also listed below (page 4) along with notes on responses you might anticipate from students. Teachers presenting through distance education platforms may opt to use the do-now activity as a writing exercise instead.

Part 2: Slideshow / Video (5 - 10 minutes)

The slideshow highlights rapid developments in genetic technologies. The accompanying explanatory notes for the slideshow are below (pages 5-7). In addition, we have recorded a video of this slideshow for easy use in a distance learning setting. The PowerPoint file as well as the video can be found on our website along with this lesson.

Extending the Lesson

Students can reflect, in writing or in a discussion, on why they might or might not be interested in genetic analysis. They can cite evidence from the slideshow, from *The Gene*, or from some of the links embedded in this lesson. We have also provided a writing or discussion prompt for teachers to use, along with some information for teachers to help guide that discussion (page 8).

Additional resources for teachers

Amy Harmon's "DNA Age" series in the *New York Times* has news, analysis and video clips related to personal genetics. While some of the science has progressed, the issues Harmon examines are still timely.

The Hastings Center is an independent, nonpartisan and nonprofit bioethics research institute with many resources and articles about ethics, genetics and society.

Related pgEd lesson plans

- This module was adapted from pgEd's lesson, Introduction to Personal Genetics.

- pgEd offers a full lesson on Consumer Genetics, which delves deeper into this topic and includes an activity for setting up a classroom debate on this topic.

DO NOW EXERCISE: TEACHER NOTES

What are the potential health benefits to knowing about your genetic makeup?

You can expect students to answer this question with ideas related to taking steps to prevent or seek early treatment for a potential health condition. You may suggest that even if students learned of conditions without a current treatment, they could choose to become active in clinical trials or advocacy.

What are the possible downsides to knowing about it?

Students frequently raise concerns related to privacy and discrimination, as well as the emotional burden of learning about one's genetic information. This can be an opportunity to tell students about the Genetic Information Non-discrimination Act (GINA), a US federal law that forbids the use of genetic information to discriminate against people in the areas of employment and health insurance.

> ***Read more:*** pgEd's lesson Privacy Protections for Genetic Information: Meet GINA focuses on the protections extended by GINA.

Students often think that genetics will give them definitive, life-altering answers, including a specific cause of death. It is important to clarify these misconceptions and instead emphasize the concept of genetic complexity. Remind students that genes are part of a much more complex story in concert with the environment, and scientists still have much to learn about the links between a person's genetic makeup, environment, and health.

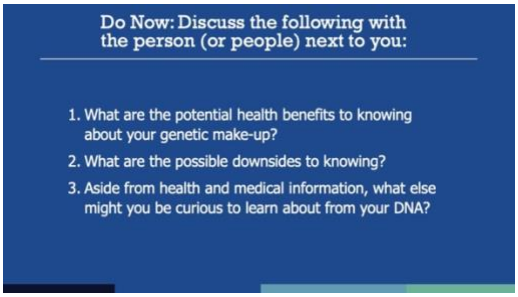
Aside from health and medical information, what else might you be curious to learn from your DNA?

Students frequently mention learning about their ancestry and finding biological relatives. Many students will be familiar with the concept of ancestry testing, and as time goes on, some may have direct experience with it. Students who are adopted or living apart from biological relatives often will specifically mention these possibilities and may be interested in reading any one of the hundreds of news stories about people's direct experiences with using ancestry tools to locate biological family connections.

SLIDESHOW NOTES

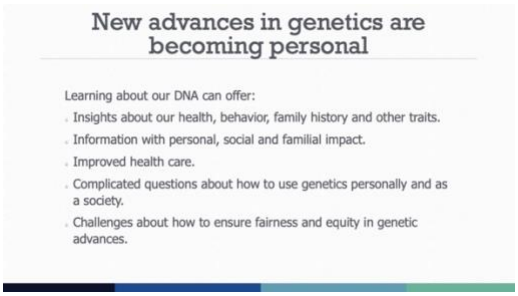
The slideshow as well as a recorded version of it are located on our website along with this lesson, and accompanying explanatory notes for the slideshow are below. The main idea of each slide is in bold along with text that summarizes the story presented in each slide. The notes provide additional context to aid in answering student questions and references for teachers interested to delve deeper into these topics.

Slide 2: Do Now exercise



The questions for this "Do Now" activity will help students begin to consider the topics covered in the lesson. Detailed notes for this slide are in the section above.

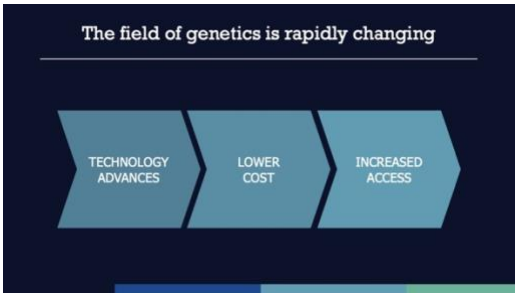
Slide 3



This slide introduces students to the field of personal genetics, highlighting the potential personal and societal impacts of widespread DNA analysis. It sets the stage for the upcoming discussions about developments in genetic technologies and ancestry testing. *In this case, "personal genetics" is used as a broad term to capture the many choices that individuals may face as healthcare consumers and as members of society.*

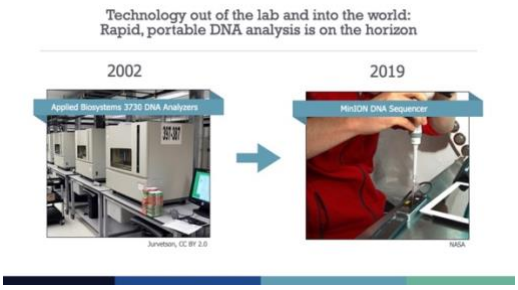
While learning about your DNA is highly personal, the ability to do so raises many questions about how we as a society should handle access to genetic information. For example, if you learn about your risk for Alzheimer's disease, this may raise concerns for your biological relatives, who might not want to know anything about their risk for Alzheimer's disease. Might easy access to genetic predispositions affect dating relationships? Should insurance companies be able to know information about your DNA? Would everyone have equal access to personal genetic information and the benefits of new genetic technologies? These are a few of the issues that must be considered as genetic technologies enter societal use.

Slide 4



Technological advances that have lowered the cost of genome analysis are paving the way for people to have greater access to genetic information. This slide sets up the main ideas in slides 5-6, which provide examples of rapid advances in technology that are changing how readily people can access genetic information. For example, when the Human Genome Project was "completed" in 2003, it cost 3 billion dollars and took about 13 years. (Note: though the project ended, nearly two decades later in 2020, there remain several sections of the genome that have not been mapped.) In 2020, sequencing a human genome can cost as little as $300-$1,000 US and can be done in a matter of days, sometimes less. Additional time and expense may be required for interpreting the genome sequences (i.e., analyzing the sequences to arrive at clinically useful information or predictions) as well as consulting with medical professionals (e.g., physicians, genetic counselors).

Slide 5

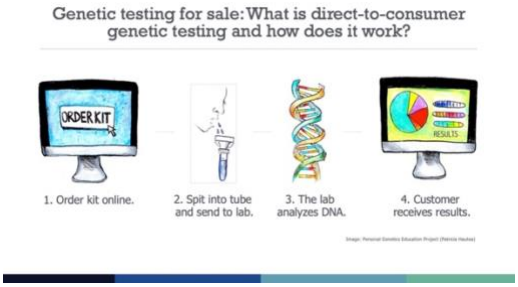


The machines that sequence DNA are becoming smaller and more accessible. The first sequence of the human genome was achieved with hundreds of sequencing machines working for years. Now a single machine can sequence a full human genome in a matter of days. To make the technology more accessible, sequencing machines are now smaller and more affordable. For example, some companies (e.g., Genapsys and Oxford Nanopore) are developing DNA analysis machines, capable of providing a partial analysis (rather than a full genome sequence), that are the size of a loaf of bread, a bar of soap, or a USB drive.

Easily transported, handheld genome sequencers can be useful in emergency medical situations, whether it be a rapid diagnosis in an emergency room or to track the spread of a virus (such as the recent outbreaks of Ebola and SARS-CoV2, which is the virus responsible for COVID-19).

> ***Read more:*** For more on genetics and Ebola, see the NIH's article here. For scientific updates about COVID-19, see NIH's resources here.

Slide 6



Genetic testing kits are for sale online and in pharmacies and department stores in the US. These so-called "direct-to-consumer" (DTC) tests from for-profit companies offer a range of services from ancestry and health risk information to information about other personal traits. Some of these can be purchased without a doctor's request or participation, while others require consulting with a doctor or genetic counselor about results. Samples are often collected in the form of spit in tubes, which are then analyzed in a lab, and results are returned to customers by logging onto a website. Some of these products look at areas of the genome that commonly differ between individuals, known as single nucleotide polymorphisms (SNPs). A typical SNP test from a DTC company might analyze 0.05% of a person's genome, but decreasing costs means some companies are now offering more comprehensive analyses by reading the full sequences of specific genes, or even providing a full genome sequence. These products may provide information about risk for cancer or other conditions, or carrier status for genetic variants that, if inherited from both parents, can cause serious diseases in their children. Other tests offer an analysis of your possible ancestry or may provide interesting but not particularly consequential information about your traits, such as whether or not you're likely to sneeze when in bright sunlight or if you are likely to enjoy the taste of cilantro.

There has been much debate over DTC genetic testing since it first emerged in the early 2000s. These testing kits have made personal genetic information more accessible than ever before and millions of individuals have sent in their DNA samples seeking information about their traits, ancestry, and health. While many people are excited to learn about their genetic make-up and believe they have the right to know whatever information may be in their DNA, others are concerned about the safety of providing individuals with genetic health information without the supervision of a healthcare professional. Worries persist that individuals will misinterpret results, feel a false sense of security, or make medical decisions based on the limited information provided by these tests. Concerns about the safety and accuracy of these tests have also led some federal and state regulators to push for them to be considered medical devices and subject to extensive rules about safety and quality.

EXTENDING THE LESSON: WRITING PROMPT & TEACHER NOTES

Ask students whether they agree or disagree with the following statement. Why or Why not?

Parents should be able to find out whatever they would like about their children's DNA, whether it be about ancestry or health, before they turn 18.

This statement is intentionally broad to encourage students to consider how much information could be available to parents through genetic testing. There is an ongoing debate about how much parents should find out about their child's DNA before age 18, when a child becomes a legal adult. Many students may have already had their DNA assessed in some way via prenatal genetic screening or newborn screening. While genetic testing can help to predict, diagnose or treat certain diseases, it may uncover genetic risks for disease later in life or other information that might affect how a child is raised. Many people are concerned about the availability of this information before the child is able to give informed consent. Should testing be limited to only look for a specific subset of genetic markers, such as those for diseases with childhood onset? If genome sequencing is performed, should only specific types of information be provided to parents? Is ancestry information considered "sensitive" or not?

In the realm of medical genetic testing, the American College of Medical Genetics and Genomics (ACMG) offers guidelines regarding the reporting of incidental, or secondary, findings when performing genome sequencing. It recommends doctors only tell patients about findings of specific genetic variants that are associated with serious disorders that have practical things that can be done to mitigate the associated risk. Patients are allowed to opt-out of receiving these results during pre-testing counselling.

> ***Read more:*** More on the ACMG's recommendations, which generated much debate and discussion, can be found here: "Considering ACMG's practice guidelines for incidental genomic findings" and "ACMG SF 2.0".