



## LESSON PLAN & TEACHER'S GUIDE

# Introduction to Personal Genetics

### Aim

How might new advances in personal genetics impact our lives, our medical decisions and society?

### Guiding questions

- How would you decide whether or not to get your genome analyzed?
- How might genome analysis change healthcare?
- What are the benefits and concerns for you and other stakeholders?

### Learning objectives

After completing this lesson, students will be able to:

- Define what is a personal genome sequence.
- Grasp the rapid rate at which sequencing technology is developing.
- Analyze why people would or would not want to know about their genome.
- Place genetic discoveries within the context of particular historical and current events.

### Materials

Projector or Smartboard, laptop, paper, tape, handouts.

### Time/classroom implementation

We have organized this lesson plan into discrete segments (listed below), each focusing on a major theme in personal genetics. We hope this model gives teachers the flexibility

to teach some or all of the segments depending on your time and classroom needs. Teachers may choose to break the slideshow into smaller pieces to delve deeper into each topic and classroom dialogue.

- Part 1:** Student overview of the emerging field of personal genetics (pre-reading, vocabulary, do now activity, slides 2-7)
- Part 2:** Genetics and health (slides 8-14)
- Part 3:** Genetics and ancestry (slides 15-18)
- Part 4:** Societal implications of genetics (slides 19-23)
- Part 5:** Classroom discussion (Four corners activity, slides 24-32)
- Part 6:** Additional assessments and student handouts

## Standards alignment

### Common Core Standards

[CCSS.ELA-LITERACY.RH.11-12.1](#) Cite specific textual evidence to support analysis of primary and secondary sources, connecting insights gained from specific details to an understanding of the text as a whole.

[CCSS.ELA-LITERACY.RST.11-12.2](#) Determine the central idea or conclusions of a text; summarize complex concepts, processes, or information presented in a text by paraphrasing them in simpler but still accurate terms.

[CCSS.ELA-LITERACY.RST.11-12.7](#) Integrate and evaluate multiple sources of information presented in diverse formats and media (e.g., quantitative data, video, multimedia) in order to address a question or solve a problem.

### Next Generation Science Standards

*This pgEd lesson integrates some of the NGSS practices and cross cutting concepts associated with the following disciplinary core ideas. The relevant portion of each disciplinary core idea is written out below.*

#### [HS-LS1 From Molecules to Organisms: Structures and Processes](#)

##### *LS1.A: Structure and Function*

- Systems of specialized cells within organisms help them perform the essential functions of life.
- All cells contain genetic information in the form of DNA molecules. Genes are regions in the DNA that contain the instructions that code for the formation of proteins.

##### *LS1.B: Growth and Development of Organisms*

- The organism begins as a single cell (fertilized egg) that divides successively to produce many cells, with each parent cell passing identical genetic material (two variants of each chromosome pair) to both daughter cells. Cellular division and differentiation produce and maintain a complex organism, composed of systems of tissues and organs that work together to meet the needs of the whole organism.

## HS-LS3: Inheritance and Variation of Traits

### *LS3.A: Inheritance of Traits*

- Each chromosome consists of a single very long DNA molecule, and each gene on the chromosome is a particular segment of that DNA. The instructions for forming species' characteristics are carried in DNA. All cells in an organism have the same genetic content, but the genes used (expressed) by the cell may be regulated in different ways. Not all DNA codes for a protein; some segments of DNA are involved in regulatory or structural functions, and some have no as-yet known function.

### *LS3.B: Variation of Traits*

- Although DNA replication is tightly regulated and remarkably accurate, errors do occur and result in mutations, which are also a source of genetic variation. Environmental factors can also cause mutations in genes.
- Environmental factors also affect expression of traits, and hence affect the probability of occurrences of traits in a population. Thus the variation and distribution of traits observed depends on both genetic and environmental factors.

## **Background information**

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 and help extend people's lives. As the cost of genetic analysis decreases and research advances, it is becoming increasingly possible to include a person's genetic make-up in the repertoire of tools that inform their healthcare. The growing field of personal genetics is at the intersection of science and society; it is both an exploration into the complex interactions through which our genes and our environment influence our physical, mental and behavioral states, as well as an on-going conversation on the meaning for individuals and society.

Today's high school and college-age students will become independent healthcare consumers in a time when genetic technologies are increasingly used in medical care, promising the ability to analyze one's genetic risks and tailor medicines and interventions to the individual. At the same time, health disparities and inequitable access to medical services prevent the potential benefits of genetics from being realized by everyone.

This lesson introduces students to recent advances in genetics and medicine, DNA ancestry testing, and reasons people may choose to learn about their genetic make-up. Students will be exposed to the societal issues and ethical challenges individuals may face regarding the use of this technology. This lesson aims to (1) put these developments in genetics into a broader social context, with a particular emphasis on persistent health disparities, and (2) explore the differences of opinion regarding how genetic information should be used personally and in society. Broken into chapters, teachers can choose some or all of the topics to introduce to students.

## Interdisciplinary connections

Students do not need to have a background in genetics, as this lesson focuses mainly on social and ethical issues related to genetic testing. The material presented crosses into multiple subjects, including biology, health, social studies, law, anthropology and psychology. If you have not done a full unit on genetics, you might first explain to students some fundamental concepts in genetics, specifically that (1) traits can be passed down from parent to child through their genes and (2) complex human traits are often the result of genes in combination with environmental factors. Students can learn more at the award-winning website from the [Genetic Science Learning Center](http://learn.genetics.utah.edu/content/inheritance/) at the University of Utah. In particular, we recommend that students watch the “What is heredity?” and “What is a trait?” videos found at: <http://learn.genetics.utah.edu/content/inheritance/>.

## Foundational concept

One important theme to weave through 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. By analyzing the genome sequences and characteristics of many people, personal genome sequencing has the potential to link traits to certain genes. The most obvious benefit of these analyses will be to better understand the interplay of nature and nurture in known diseases, with the hope that they would lead to better treatments, cures, preventative measures and healthier generations of children.

Identical siblings, such as twins or triplets, arise from a single embryo, so their DNA sequences are virtually the same, with the exception of differences that arise over time, for example by the process of mutation.

## Guide for using the lesson plan

This lesson includes a do now activity, a slideshow, and a four-corners activity.

The slideshow is located at <http://www.pged.org/lesson-plans/> along with this lesson. Accompanying explanatory notes for the slideshow are provided in the sections below. You may want students to take notes to help evaluate their understanding, particularly

if you will be using additional [pgEd lesson plans](#), and students will later participate in a debate and/or writing activity. One excellent method for note-taking is [Two Column Note Taking](#). A sample graphic organizer is included on [page 29](#).

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## Additional resources for teachers

1. 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.
2. The [Hastings Center](#) is an independent, nonpartisan and nonprofit bioethics research institute with many resources and articles about ethics, genetics and society.
3. Scientific background for some of the [ancestry concepts](#).

## Related pgEd lesson plans

[Consumer genetics](#)

[Personalized medicine](#)

[Genetics, jobs and your rights](#)

[Genetics and reproduction](#)

[Genome editing and CRISPR](#)

[Athletics and genetics](#)

[Protecting athletes with genetic conditions: Sickle cell trait](#)

[DNA, crime and law enforcement](#)

## Introduction to Personal Genetics

### Part 1: OVERVIEW FOR STUDENTS

#### Reading for students

To give students a sense of the “big picture” in personal genetics, we recommend Carl Zimmer’s “[Game of Genomes](#)” series in *STAT News*. It is a lengthy series, and teachers may want to recommend reading only part 1 or 2 in advance of this lesson. While the regulatory landscape for the sale of direct-to-consumer genetic tests evolves, and the technology and costs are always changing, the issues explored in this series related to privacy, health, identity and the question “what do I want to know about my DNA?” remain very current.

#### Vocabulary

There are a number of vocabulary words that may be unfamiliar to students. You can provide a vocabulary list, or have students look up words themselves.

**Gene** – A basic functional unit of genetic information in our DNA.

**Genetic test** – A procedure that gives some information about the genetic make-up of an individual.

**Genetic variant** – One of several possible DNA sequences at a particular location in the genome.

**Genome** – An individual’s full set of genetic information, including all genes as well as other sections of DNA that may regulate when genes are turned on or off.

#### Do Now exercise (7 minutes)

Have students answer the following questions individually or in pairs and then share their answers in a brief classroom discussion. These questions are on Slide 3 in the slideshow.

1. What are the potential health benefits to knowing about your genetic make-up?
2. What are the possible downsides to knowing?
3. Aside from health and medical information, what else might you be curious to learn about from your DNA?

You can expect students to answer question one with ideas related to taking steps to prevent or seek early treatment for a potential health condition. You may suggest that

even if they learned of conditions without a current treatment, they could choose to become active in clinical trials or advocacy.

For question two, 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 Nondiscrimination Act](#) (GINA), a federal law in the United States which forbids the use of genetic information to discriminate against people in the areas of employment and health insurance. pgEd's lesson "[Genetics, Jobs and Your Rights](#)" focuses on the protections extended by GINA.

Students often suggest genetics will give them definitive life-altering answers, often 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 make-up, their environment, and their health.

For the third question, students frequently mention learning about their ancestry and finding biological relatives.

## Interactive slideshow (Part 1)

Part 1 of the slideshow (located at <http://www.pged.org/lesson-plans/>) highlights rapid developments in genetic technologies. The main idea of each slide is in bold, and accompanying notes summarize 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. Please reference the teacher's guide ([page 1](#)) for key points to emphasize with students.

## Slideshow Notes

### Slide 2

**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 applications for healthcare, ancestry explorations, and the criminal justice system. *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 this lesson will explore.

### Slide 3

**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 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, over a decade later in 2019, there remain several sections of the genome that have not been mapped.) In 2019, [sequencing a human genome](#) can cost as little as \$700-\$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.

One hope is that 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 Ebola, as was done during a recent outbreak). For more on genetics and Ebola, see the NIH's article [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 collected usually 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).

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.

See pgEd's [lesson](#) on "Consumer Genetics" for more information.

## Slide 7

**Technological and scientific advances in genetics are impacting healthcare and medicine, adding exciting and complicated layers to concepts of identity and history, and having an impact on public life – particularly, the criminal justice system.** These topics will be addressed in subsequent slides (see Parts 2-4 below).

## Introduction to Personal Genetics

### Part 2: GENETICS AND HEALTH

#### Interactive slideshow (Part 2)

Part 2 of the slideshow (located at <http://www.pged.org/lesson-plans/>) gives examples of how advances in genetics have led to medical breakthroughs and potential benefits for health. The main idea of each slide is in bold, and accompanying notes summarize 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. Please reference the teacher's guide (page 1) for key points to emphasize with students.

#### Slideshow notes

##### Slides 8-9

**One major use of genetic testing is to determine whether an illness is genetic, with the hope of identifying a cure or treatment plan.** Others find a long-sought-after answer when visits to specialists failed to yield a diagnosis. This slide highlights an example of how families with rare genetic disorders are seeking to connect with others to share medical resources and emotional support. It is also an example of how families can take action, even when faced with a medical condition that doesn't yet have a well-established path of treatment.

This slide highlights the story of Tess Bigelow, a young girl whose genome was analyzed in search of a genetic cause for a number of complex health issues and developmental delays. The analysis revealed a change in her USP7 gene that caused it to malfunction. Disorders of the USP7 gene are very rare, so Tess's father, Bo, made a social media post in search of others children with the same genetic variant as Tess. The following day, someone who saw Bo's post connected him with a researcher studying this gene, and Bo learned that there were seven other children known to have the same condition. The Bigelow family have become activists, in addition to champions for Tess and her medical journey, seeking to find and connect families all over the world who might be affected by this genetic variant. They launched a foundation and created a community of other affected families in hopes of one day finding enough patients to populate a research study. The Bigelow family tells their story [here](#).

## Slide 10

**One reason people seek genetic testing is to identify future health risks.** In May 2013, actor Angelina Jolie revealed that she had undergone a double mastectomy because she carries a mutation in the BRCA1 gene that greatly increases her risk of breast and ovarian cancer. Some believe a preemptive double mastectomy was a dramatic and potentially unnecessary choice since Jolie neither had cancer nor knew with certainty that she ever would, but according to Jolie's op-ed "[My Medical Choice](#)" in the New York Times, her chances of developing breast cancer dropped significantly as a result of the surgery. This is a great way to engage students with a real-life example of a well-known person who used the results of a genetic test to inform a major medical decision aimed to reduce her risk of disease. Jolie's advocacy, alongside many others, includes the goal of making genetic testing more accessible to all who want it. To learn more about breast cancer, BRCA1 and other risk factors, see the following link from the American Cancer Society: [www.cancer.org/cancer/breastcancer/detailedguide/breast-cancer-risk-factors](http://www.cancer.org/cancer/breastcancer/detailedguide/breast-cancer-risk-factors)

## Slide 11

**People may learn how their DNA affects their response to certain drugs.**

Certain genes control the rate at which a person's body metabolizes a drug, and this can affect the dosage or whether the medicine is prescribed at all. A specific example is codeine, an opioid drug frequently prescribed for pain. Codeine is metabolized by the body into morphine with the help of an enzyme called CYP2D6. Variants in the CYP2D6 gene can dramatically affect how quickly an individual metabolizes this drug, and it can have serious, sometimes lethal, consequences if the dosage is not calibrated correctly. The FDA has cleared genetic tests to determine if a person is a rapid metabolizer (including a DTC test that was approved in November 2018), but as of 2019 they are not routinely used in the clinic.

For more information, read "[Why Codeine is Dangerous in Some Kids after Tonsillectomy](#)," by David Kroll, February 2013, Forbes. From the article:

*"...A small percentage of people are born with a single-letter change in their DNA for this enzyme that causes the codeine-to-morphine reaction go faster. These people are called ultra-rapid metabolizers.*

*Being an ultra-rapid metabolizer sounds like a good thing. But if you are and take codeine, you convert much more of it to morphine than in most people. If you're a kid with breathing problems from swollen tonsils and/or adenoids and are an ultra-rapid metabolizer, and are getting codeine for post-operative pain, the usual therapeutic dose is potentially lethal."*

The main point to emphasize to students is that the goal is to use information about a person's genetic make-up to identify medications that will be effective with minimal side effects, reduce negative health effects and lower costs. Another example teachers may want to introduce to their students is the risk of taking hormonal birth control, such as "the pill," for people who have a family history of a particular blood clotting condition caused by the genetic variant called Factor V Leiden. More [here](#) from the Mayo Clinic. For more on genetics and medicine, see pgEd's lesson on "[Personalized Medicine](#)."

## Slide 12

**DNA testing can be used to learn about future generations.** It is now possible to analyze DNA from a fetus through a test performed on a blood sample from the pregnant person. This test, known as non-invasive prenatal testing (NIPT), makes information about the genetic make-up of a fetus available at earlier stages of pregnancy than ever before.

*In depth: How does NIPT work? All of us have small fragments of DNA, released from cells in our body, that circulate in our bloodstream. When a person is pregnant, a small fraction of this cell-free DNA originates from cells of the placenta, an organ formed by embryonic cells that connects the developing embryo (and later fetus) to the uterus and facilitates the exchange of gases, nutrients, and waste products. By obtaining a blood sample from the arm of the pregnant person, physicians and researchers can analyze its cell-free DNA to reveal information about the developing fetus. For this reason, this test can also be referred to as "cell-free fetal DNA testing."*

pgEd uses the term "pregnant person" in recognition that not everyone who enters pregnancy would become a "mother" (e.g., in case of surrogacy) or identifies as a woman.

## Slide 13

**Genetic testing can be used to learn about the genetic makeup of embryos created by in vitro fertilization (IVF).** First, teachers may need to introduce their students to IVF, a process in which sperm and eggs are fertilized outside of the body in a petri dish, and then implanted to the uterus with the hope of an ensuing pregnancy. The first baby was born via IVF in 1978. People may use IVF in a variety of circumstances, including fertility problems or non-traditional family structures.

In some cases, a genetic testing technique called preimplantation genetic diagnosis (PGD) is performed prior to transferring embryos to the uterus. For PGD, one or more cells are removed from embryos created by IVF, and DNA from these cells is then tested for various genetic markers. First performed in 1990, PGD is often used in an effort to avoid genetic disorders, including deadly or debilitating childhood diseases

such as Tay-Sachs and cystic fibrosis, as well as adult-onset diseases such as Huntington's disease and breast cancer. In some parts of the world, including the United States, PGD can also be used for non-medical sex selection based on parental preference.

Access to IVF and PGD remains quite limited due to cost. In 2019, the average cost of IVF and PGD, combined, is \$20,000-30,000 US. While this procedure is covered by public health care systems in some other countries, fewer than a third of US states (covering just less than half of the US population) mandate insurance coverage for infertility treatments. For many people without health insurance, the procedure remains a distant prospect.

## Slide 14

**The case of Layla Richards symbolizes the potential promise of genome engineering for treating diseases.** Diagnosed at 14 weeks old with leukemia, a type of cancer that affects blood and bone marrow, Layla Richards was 11 months old when all conventional treatments had failed. Layla became the first child to be treated for leukemia via donated immune cells that were genetically engineered specifically for her body and type of cancer - a kind of treatment called immunotherapy. The cells, called CAR-T cells, were engineered to attack Layla's cancer cells. The cells were also altered to ensure Layla's immune system would not perceive them as dangerous and reject them. The transplant was a success, and as of the most recent report in early 2017, Layla remained cancer-free.

["A Groundbreaking Gene-Editing Therapy Eliminated Cancer in Two Infants,"](#) by Kristen Brown, January 2017, *Gizmodo*.

## Introduction to Personal Genetics

### Part 3: GENETICS & ANCESTRY

#### Interactive slideshow (Part 3)

Part 3 of the slideshow (located at <http://www.pged.org/lesson-plans/>) gives examples of how advances in genetics are impacting people's explorations of their ancestry. The main idea of each slide is in bold, and accompanying notes summarize 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. Please reference the teacher's guide (page 1) for key points to emphasize with students. Mid-way through the slideshow, pause to show either one of the short videos "[Black People Get Their DNA Tested](#)" or "[Who do we think we are?](#)", which are also linked in the slides.

#### Slideshow notes

##### Slide 15

**Ancestry testing is another dimension of personal genetics.** In 2019, DNA ancestry tests often cost \$100 US or less. For some, genetics can add a layer of information onto decades or centuries of records of family history. For others, whether because of adoption, migration, or other family circumstances such as donor-assisted conception, might be seeking unknown relatives from all over the world. For example, because of the history of slavery in the US, many people of African ancestry do not know in what part of Africa their ancestors lived. DNA testing may provide some information towards filling in these gaps and may allow people to feel more connected to a past of which they were previously unaware. On the other hand, DNA testing can also complicate or even contradict a person's sense of identity if results are unexpected or unwelcome. At the same time, some ethnic groups, particularly many Indigenous American nations, place a greater emphasis on cultural belonging and familial lineage rather than genetic ancestry when considering questions of ethnic identity.

##### Slide 16

**These maps illustrate the point that while some people's ancestry can be seen via a world map looking at human migration patterns in the ancient past, others will see the history of their families more clearly in a map of the slave trade.**

Using a combination of the archaeological record and DNA analysis, scientists have been able to track human movements out of Africa, believed to have started around 60,000-70,000 years ago, to eventually most areas of the world (except for Antarctica). The top-left image illustrates current understanding of ancient human migration across the globe, showing the approximate date when a particular island or part of a continent is believed to have been first inhabited by humans. Importantly, this map doesn't show the subsequent migration of different populations to areas that were already inhabited (e.g., the expansion of peoples speaking Indo-European languages from southern Russia into Europe and India, or the migration of Bantu-speaking peoples from central Africa into southern Africa, both of which occurred within the past few thousand years).

The second map illustrates the most common transatlantic slave trade routes during the 17<sup>th</sup>-19<sup>th</sup> centuries. During this time, 10-15 million people were forcibly moved from one part of the world to another. Today, there are many more millions of descendants of this slave trade whose genealogical and cultural history have been impacted by this forced migration. Taken together, these maps illustrate some of the many historical events that have shaped the genetic ancestry of populations across the world today.

## Slide 17

**This slide illustrates the complex relationship between nationality, ethnicity, ancestry, and physical traits seen in countries formed as a result of European colonization, such as Mexico and the US.** Such countries are not ethnically-uniform, but are instead multicultural and multiethnic countries with varied percentages of ancestral origins in their populations. National/ethnic identities such as "Mexican" can encompass dozens of languages and cultures, a mix of ancestries (including African, Indigenous American, European and/or Asian ancestry), and a wide diversity of traits. The variation in traits such as skin color is seen on this slide. In the US, many people of Mexican and other Latin American origins may identify or be identified as "Latino." The term "Latino" came into use largely as a category in the US Census; it does not easily map onto common notions of race and geography and is not broadly used across the globe.

## Slide 18

**The topics covered in slides 15-17 are brought to life in two videos in which young people are learning about their genetic ancestry via direct-to-consumer genetic tests.** Both videos include people reflecting on how their results from a DNA ancestry test might fill in important pieces of their personal story, give them a sense of belonging, or differ from their family stories and concepts of ethnicity and identity. Also, both videos mention the history of colonization and slavery, and how painful that can be within families. The two videos both reinforce the idea that people are more alike than different from a genetic perspective.

The two videos differ in several ways. The video "Black People Get Their DNA Tested" from *BuzzFeed News* specifically mentions they are using a 23andMe product. (Important note: While we have included this video, pgEd does not recommend for or against any specific product.) We also see the interactions between family members as they process the information they've received. Lastly, this video contains some images about current political matters in the US - including immigration and the Black Lives Matter movement - through the lens of how genetic testing could remind people of our common humanity. In the NIH-produced video, "Who do we think we are?", you will see a more classroom-style approach, with more context about how DNA tests work, the history of human migration, and a discussion about the risks related to discovering unexpected information.

After watching either video, remind students that ancestry results can vary widely between companies, and rather than being definitive, are often a reflection of whose DNA is included in the database and how these "reference" samples are divided up into ethnicities or geographic regions. Many ancestry databases are over-represented with samples from people of European descent, leading to less precise results for people of non-European descent. pgEd advises teachers to watch both videos and decide which one (if not both of them) will meet your needs.

["Black People Get Their DNA Tested"](#) by *Buzzfeed*

["Who do we think we are?"](#) produced by 42o North Media for NHGRI

## Introduction to Personal Genetics

### Part 4: SOCIETAL IMPLICATIONS OF GENETICS

#### Interactive slideshow

Part 4 of the slideshow (located at <http://www.pged.org/lesson-plans/>) puts new developments in genetics into a historical and societal context. The main idea of each slide is in bold, and accompanying notes summarize 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. Please reference the teacher's guide (page 1) for key points to emphasize with students.

#### Slideshow notes

##### Slide 19

**Personal genetics is also impacting law enforcement and privacy practices, and is a microcosm of larger debates about trust, access, and transparency in science.** The following slides provide brief examples of these ideas, and highlight 1) past failures in research ethics, 2) new law enforcement techniques, 3) the struggle for healthcare access, and 4) new models of community participation in genetics.

##### Slide 20

**In the US, suspects are being identified not just through criminal databases - but also genealogy databases designed for researchers and hobbyists.** After failing to find a match in the government-created databases, investigators in "The Golden State Killer" case uploaded what they believed to be the notorious rapist and murderer's DNA to an open-source genealogy database, called GEDmatch. GEDmatch is a privately-created database that welcomes people to upload their DNA analysis from private companies like 23andMe or Ancestry.com, in the hopes of building a large community for people seeking familial connections. Law enforcement found a genetic connection in the database – a distant cousin of the suspected killer. Using genealogical research to construct a family tree, investigators narrowed down the possible suspects and, with additional DNA testing, an arrest was made. Joseph James DeAngelo is under arrest for the crimes and is awaiting trial as of June 2019.

This case is an example of how quickly a new technique can take hold. Though GEDmatch was not developed to be a legal tool, in the months after the arrest of the suspected "Golden State Killer", law enforcement agencies used the database to make

arrests in several other “cold cases.” Some people have reacted positively to this news by saying that any and all methods are justified in the pursuit of solving crimes. Others have voiced concern regarding the fact that if even one biological relative uploads their DNA to a genealogy database like GEDmatch, then some of their shared DNA is also part of a system that is now being used for law enforcement reasons. Additionally, decisions about law enforcement access to the GEDmatch database is largely in the hands of two private citizens who founded the organization.

In May 2019, GEDmatch changed its terms of service so that DNA profiles are now by default opted out of use for law enforcement investigations. Users are able to opt-in if they wish to do so. As of June 2019, only 5% of the 1 million members of GEDmatch have opted-in. In December 2019 it was announced that GEDmatch was taken over by the forensic genomics firm Verogen.

[“How a Tiny Database Became the Police’s Go-To Genealogy Database”](#) by Sarah Zhang, June 2018, *The Atlantic*.

## Slide 21

**This slide highlights that fact that many people in the US, across ethnicities, cannot access the healthcare services they need.** This remains a pervasive problem, and one that should be included in the discussion of how the potential of personal genetics to improve health remains out of reach for many until these underlying issues of access and economics are addressed.

## Slide 22

**The Tuskegee Syphilis Study serves to illustrate past breaches of trust in medical research.** The Tuskegee Syphilis Study, a study of the natural course of untreated syphilis, was run by the US Public Health Service from 1932-1972. Over 600 disenfranchised African American men were enrolled in the study and were to receive free healthcare and burial insurance. Many of the men were not told they were part of a study, nor were they informed about the risks or advised that they could leave the study. Even though penicillin was the standard drug given to treat syphilis by 1947, it was withheld from the men in the study. The study came to an end in 1972 after it was reported in the press. It is considered one of the most egregious human experiments in the US, because the men were not properly consented and treatment was withheld. Many of the men died, 40 of their wives were infected as well, and 19 children were born with congenital syphilis. President Clinton formally apologized to the survivors in 1997. The *Belmont Report*, produced by a national commission that was formed in part to reckon with how the Tuskegee study was ever allowed to happen, altered informed consent protocols and oversight of research involving human subjects. Read more: [“How the Public Learned about the Tuskegee Syphilis Study”](#) in *Time*.

Other examples teachers might wish to expand on include the American eugenics movement (full pgEd lesson plan [here](#)), the story of [Henrietta Lacks](#), or the [abuses by Dr. Marion Sims](#), considered a pioneer in women's reproductive medicine but whose work relied on brutally painful and repetitive surgeries on enslaved women during the 1850s in the US.

These histories, which have created distrust and fear of biomedical researchers, are one reason for the [much lower](#) rate of participation in clinical research or drug trials among ethnic minority communities than white communities. Researchers continue to strive to increase the diversity of research participants by increasing awareness and building trust with these communities. For more, see:

["To advance medicine's future, the NIH tries to win the trust of communities mistreated in the past,"](#) by Lev Facher, September 2017, *STAT News*.

## Slide 23

**Collaboration between researchers and communities that emphasizes trust, access and transparency is an emerging model in genetic research.** An example of this approach to genetic research is being implemented by the Diné (also known as the Navajo Nation). Navajo leaders have proposed lifting the ban on genetic research that the nation had put in place since 2002 so their community can benefit from advances in genetics. Tribal leaders are devising a [new model of participation](#) in research to avoid potentially unwanted or inappropriate use of their DNA. In this model, Navajo Nation would have the power to approve or reject research studies and retain control over their DNA samples. They are also conducting public meetings and hearings to gather input.

Another example is the formation of an advisory panel at the National Institutes of Health giving the descendants of Henrietta Lacks a voice in the use of their relative's genetic material in research. A collaborative approach is also being used in Massachusetts, New Zealand and sub-Saharan Africa where genetic technologies are being considered to solve environmental issues with insect-borne disease and invasive species. Projects such as "[Responsive Science](#)" are creating new models of collaboration with communities, placing the wishes of the people living in the impacted areas in the center of the discussions about if and how a project will proceed.

## Introduction to Personal Genetics

### Part 5: CLASSROOM DISCUSSION

#### Four Corners activity (15-25 minutes)

Students will hear several statements and move to labeled corners of the room based on the degree to which they agree or disagree with each statement. They will share and discuss their opinions with the class.

##### Step 1: Preparation

Before students enter the classroom, label the four corners of the room with signs reading: *strongly agree*, *agree*, *disagree* and *strongly disagree*. Depending on your classroom, you could also use a straight line across the room and ask students to place themselves on the continuum between strongly agree and strongly disagree.

Remind students that new advances in genetics and genome sequencing offer great hope for medicine and health; at the same time, there are questions to consider on a personal and societal level. The statements below offer students a chance to express their opinion and discuss some of the issues based on what they learned in the slideshow.

##### Step 2: Introducing statements

We have provided eight statements, included on Slides 25-32 of the slideshow and listed below on pages 22-25 along with teacher notes. Teachers can choose which slides will work best for their classroom, and teachers report to pgEd that using just 4-6 of them can advance the conversation and get students thinking in new ways. Before beginning the discussion, remind students about norms for having a respectful, open discussion of ideas.

Read each statement and have students move to the corner of the room corresponding with whether they strongly agree, agree, disagree or strongly disagree with the statement. (Displaying the statements on the slides will help students understand the statement and prevent the teacher from having to read it several times.) After students are in their places, have them discuss in their groups why they agree/disagree. Then, ask for volunteers to explain their position to the group; they should cite information from the homework article or slideshow discussion that supports their position. Encourage students to switch corners if someone presents an idea that causes them to change their opinion. After a representative from each corner has defended his or her position, you can allow students to question each other's evidence and ideas. If you have longer class periods or will use this as a pre-writing activity for either an essay or

debate, you can distribute statements and give students the opportunity to respond to them in writing before discussing.

Note: An important concept to raise throughout this discussion is that many physical, mental and behavioral traits are very difficult to predict or ensure. This is because such traits are the result of an individual's environment and lifestyle as well as an individual's genetic make-up. In addition, the genetic basis of many traits is extremely complex and beyond our current scientific understanding.

### Step three: Debriefing

You can debrief the lesson as a class discussion or individual writing assignment. Discuss or have students write about how the activity changed or reinforced their original ideas. To clarify ideas shared during the discussion, chart the main arguments on the board as a whole-class activity.

### Statements and teacher notes

***I would want to find out my likelihood of developing a disease only if there are ways to prevent or treat it.***

Note: Even in the absence of medical interventions, some people may use this information to guide lifestyle changes and financial planning or may be spurred into advocacy or participation in research. Other people feel that concerns about discrimination and the emotional impact override the benefits of having this information. Some studies have shown overall that learning about heightened genetic risk for disease does not appear to do long-term harm. To read more, see "[Learning of Risk of Alzheimer's Seems to Do No Harm](#)," by Denise Grady, July 2009, *New York Times*.

***People should be required to get counseling from a doctor or genetic counselor when they get genetic testing because they will not be able to handle the information otherwise.***

(See note for next slide.)

***People should have the right to learn whatever they want about their DNA because it is their own body.***

Note: The previous two statements show conflicting viewpoints about genetic testing, particularly in regard to direct-to-consumer (DTC) testing. As discussed in Slide 6, there has been much debate over DTC genetic testing. One question is whether genetic

testing kits should be considered medical devices, subject to the extensive rules and regulations that oversee safety and quality.

Some people opposed to DTC testing argue that people should only learn about their DNA from a doctor or genetic counselor to ensure that the results are clearly explained. People who support DTC testing often find this argument paternalistic and believe that individuals have the right to learn about their own DNA if they choose. To learn more about both of these points of view, read the following two links: "[Why You Should Think Twice About At-Home Genetic Testing](#)" (cautious about DTC testing) and "[American Medical Association: You can't look at your genome without our supervision](#)" (supportive of DTC testing).

***Parents undergoing in vitro fertilization should have the option to screen embryos for genetic variants likely to cause a serious disease.***

Note: Some prospective parents who know they carry a genetic variant that would likely cause serious disease or death in a child opt to get special testing, called preimplantation genetic diagnosis (PGD), to test embryos for that variant. Any embryos that are free of the genetic variant for which they are being tested are then considered for transfer into the uterus. Some people see this as a great advance that can reduce serious and deadly diseases, while others believe that any testing of embryos is unethical because it involves removal of one or more cells from the embryo. Another concern is that screening embryos will alter individual and societal acceptance of disability and human genetic variation. pgEd has a complete [lesson](#) on the topic of PGD and reproductive technology.

***Parents should be able to choose a child's traits, such as eye color and sex, for non-medical reasons.***

Note: Many clinics that offer PGD for medical reasons, such as to avoid X-linked diseases, also allow parents to select the sex of the child for social reasons. While the service is not widely available, at least one clinic in the US now offers eye color selection as well.

After discussing the statement, ask students to imagine in the future that it becomes possible to analyze fetal genomes to test for athletic ability or perfect pitch (the ability to identify or recreate a musical note without a reference pitch). Would they change their answer? An important concept to raise is that many physical, mental and behavioral traits are very difficult to predict or ensure. This is because such traits are the result of an individual's environment and lifestyle as well as an individual's genetic make-up. In addition, the genetic basis of many traits is extremely complex and beyond our current understanding. pgEd has a complete [lesson](#) on the topic of athletics and genetics.

***Employers should use genetic information to make hiring or firing decisions about employees.***

Note: In addition to prohibiting discrimination based on genetics when buying health insurance, The Genetic Information Nondiscrimination Act (GINA), passed by the United States Congress in 2008, prohibits employers from making hiring or firing decisions based on genetic information. Therefore, from a legal standpoint, the correct answer is simply no, employers cannot use genetic information in hiring, firing or promotion decisions. However, this statement may get students thinking about genetic traits and career choices. It raises a number of interesting questions. Can everyone be a professional athlete? Truck driver? Professor? Soldier? pgEd has a complete [lesson](#) on the topic of genetics, jobs and privacy rights. We also have a [video message](#) about GINA from former US Representative, the late Louise Slaughter, and a [5-question quiz](#) about genetic discrimination on our Map-Ed platform.

***Parents should be able to find out whatever they would like about their children's DNA before they turn 18.***

Note: 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. Teachers may want to point out that students may have already had their DNA assessed in some way via prenatal genetic 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? If genome sequencing is performed, should only specific types of information be provided to parents?

The American College of Medical Genetics and Genomics (ACMG) guidelines regarding the reporting of incidental, or secondary, findings when performing genome sequencing recommend doctors only tell patients about findings of specific genetic variants which 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.

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](#)"

***I would want to know if someone I was dating had a strong genetic predisposition to a serious disease.***

Note: The goal of this statement is to encourage students to think about how the availability of genetic information might impact our social perceptions and how private or accessible they want their information to be.

## **Additional questions for discussion**

The potential benefits of personal genetics in medicine are tremendous, but there are scientific, societal and individual challenges as well. The following four questions are included on Slide 33 and can be used to stimulate additional classroom discussion. We have included additional information for teachers below. These notes can be condensed for students or help guide classroom discussion that might result from these questions. Alternatively, these questions can be assigned for homework or used to differentiate the lesson.

### ***1. How likely will you or your doctor take action based on genetic information?***

“Take action” describes a concept that doctors often call “clinical utility.” Scientists are still learning how to interpret genomes; research is progressing quickly, but much remains to be discovered. There is no simple explanation as to how genes and environment interact to contribute to complex characteristics. Sometimes, there is a strong link between a single gene and a person’s risk for disease; however, this is not always the case. For example, scientists have uncovered over 150 genetic markers that impact a person’s susceptibility for type-2 diabetes and obesity, and it is likely that more remain to be identified. One challenge is to integrate this information with a person’s environmental factors into a personal risk assessment. One of the concerns about DTC testing is how companies calculate a person’s risk for various diseases, or that DTC testing could alarm people needlessly or provide false assurances. Furthermore, a person’s risk estimates are subject to change over time as new discoveries are made, and as a person’s genome accumulates genetic changes over their lifetime as a result of environmental factors and normal biological processes. Individuals who choose to learn about their genetic make-up should therefore be aware of the challenges in making disease risk predictions and have the interpretation of their genome reassessed periodically.

A second question is whether learning about a person’s medical condition or risk for disease is accompanied by a clear medical action for treatment or prevention. For some, genome sequencing has brought a diagnosis without a cure or has not pointed to a diagnosis at all. Others have received a much wished-for diagnosis, ending months or years of worry and speculation without any concrete information.

### ***2. How might this information impact you and your family?***

*Personal impact:* Would you want to know if you are at increased genetic risk for a disease such as cancer, for which there may be a number of steps one can take for prevention or early detection? What about Alzheimer’s disease, which currently has no effective treatment or cure? For some individuals, knowledge is power; information about their disease risks might affect how they choose to live and plan for their future. Others would prefer not to know; one reason might be that it would be impossible to go back and “unknow” this information.

*Family impact:* Since biological relatives share portions of their DNA, information that people learn about their own genetic make-up may have implications for their family members. For example, imagine a woman whose mother’s family has a history of breast cancer. Her results implicate her biological relatives – mother, siblings, children – and she will need to consider the obligations, risks, and benefits of sharing her genetic news with her relatives, or even talking with them in advance of testing.

### **3. How can we ensure access for everyone?**

*Equity of access:* How can we ensure that everyone who wants it can access their genetic information? There is general agreement that everyone, regardless of socioeconomic status, should know about the potential benefits of genome sequencing as well as the concerns, but it will take time to implement a comprehensive educational program. Can we ensure equitable access? Will payers and insurers cover the cost of genome sequencing? Will people enroll in research studies? Will prices continue to fall? Ensuring the accessibility of genome sequencing to all who are interested is a growing issue if we as a society hope to see personal genetics play an increasingly important role in healthcare.

### **4. Will people understand that our environment (healthcare, family, society, etc.) also shapes who we are?**

*The influence of factors beyond genetics:* Our physical, mental and behavioral states are the result of complex interactions between many genes in combination with our environment and our lifestyle. For example, a person may have a higher genetic risk for developing obesity or diabetes but be raised in a family or neighborhood where healthy food choices and exercise are available and feasible, which can counterbalance their genetic predisposition. Similarly, a person may have a higher genetic risk for addiction, but never try alcohol or cigarettes and avoid any likelihood of addiction. Genetic complexity is a key concept in personal genetics, and one that is explored in greater depth in several of pgEd’s lessons, including [“Athletics and genetics”](#) and [“Genes, environment and genetic complexity: Aggression in humans.”](#)

## Introduction to Personal Genetics

### Part 6: ASSESSMENTS & HANDOUTS

#### Homework assignment

Have students answer the questions below (see [page 29](#) for handout).

1. Explain some of the reasons people choose to get genetic analysis. What are some of the possible benefits? What are some of the possible disadvantages? Explain.
2. Do you think people should be able to get genetic testing directly from companies, instead of going through their doctors? Why or why not?
3. Would you consider getting genetic testing to learn more about your health? Why or why not? If yes, under what circumstances might you consider it?

#### **“Introduction to Personal Genetics” quiz answer key (see [page 30](#) for quiz)**

1. D
2. T
3. T
4. F

# Introduction to Personal Genetics

## NOTES

Name: \_\_\_\_\_ Date: \_\_\_\_\_

<b>Key Ideas:</b> What ideas are most important to remember? What new terms or concepts have been introduced:	<b>Response:</b> What questions does this information raise for you? What other ideas, events or texts does this information remind you of? Why do you think this information is important? How does this information connect to your own life? What do you think of these ideas?

## Introduction to Personal Genetics

### HOMEWORK

Answer the following questions using the information from the slideshow and/or ideas from the class discussion. Each answer should be at least one paragraph long.

1. Explain some of the reasons people choose to get genetic analysis. What are some of the possible benefits? What are some of the possible disadvantages? Explain.
2. Do you think people should be able to get genetic testing directly from companies, instead of going through their doctors? Why or why not?
3. Would you consider getting genetic testing to learn more about your health? Why or why not? If yes, under what circumstances might you consider it?

## Introduction to Personal Genetics

### QUIZ

Name: \_\_\_\_\_ Date: \_\_\_\_\_

1. People may consider having their DNA analyzed to learn about:
  - A. ancestry
  - B. elevated or lowered health risks
  - C. the genetic bases for traits including hair color, eye color and taste sensitivities
  - D. all of the above
2. Tess Bigelow used genome sequencing to discover she has a rare genetic variant, and her family is seeking other children with the same variant for a research study. T/F
3. The Genetic Information Nondiscrimination Act protects people's genetic information in the workplace. T/F
4. The cost of genome sequencing is rising and becoming more expensive. T/F