



SNAPSHOT

Introduction to Genetics and Medicine

Adapted for PBS LearningMedia in partnership with WETA for use with

KEN BURNS PRESENTS
THE GENE
AN INTIMATE HISTORY

Big Picture: Why might a healthy person choose to get genetic testing?

There are many reasons a person may choose to learn about their genetic information. For some, a difficult-to-diagnose disease might be identified thanks to genetic testing. Others may choose to learn about their genetics when they are healthy so they can use this information when making medical decisions, such as the ones described below.

Do Now: Watch [this clip](#) from the PBS documentary, *The Gene: An Intimate History*.

1) To identify future health risks



Credit: [Gage Skidmore](#),
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In May 2013, actor Angelina Jolie shared that she had surgery to remove her breast tissue. She decided to have this surgery because she learned she carries a certain version of a gene called BRCA1. We all carry this gene, but the particular version she carries greatly increased her risk of getting breast cancer and ovarian cancer. According to an article Jolie wrote in *The New York Times*, her chances of developing breast cancer dropped significantly as a result of the surgery, so this may have been a life-saving measure. However, some people believe this surgery may have been possibly unnecessary, since Jolie did not have breast cancer at the time of her surgery and might never have developed the disease. Jolie and many others want this type of genetic testing more available to anyone who might want it.

Read Jolie's article and then answer Question 1 in writing on the worksheet (page 4): "[My Medical Choice](#)," by Angelina Jolie, May 2013, *The New York Times*.

2) To learn about response to certain medications

There are genes that affect how quickly a person's body breaks down (metabolizes) certain medicines. Information about which versions of these genes a person carries can be used to decide what medicine to use or not use. This information can also be used to make decisions about how much of a medicine will be safe and effective.



A specific example is codeine, a drug from the family of medicines known as "opioids", that is often prescribed for pain. Codeine is broken down by the body into a chemical known as morphine, which is the active form of this medicine. This breakdown, or metabolism, happens in part thanks to a gene called CYP2D6. People can carry different versions of the CYP2D6 gene, which can dramatically affect how quickly they metabolize codeine. For people who carry certain versions of this gene, taking codeine can lead to serious, sometimes fatal, consequences. There are genetic tests that can be used to find out which version(s) of the CYP2D6 gene a person carries, but the practice of ordering the genetic test before prescribing the medication has not been widely adopted.

Read the excerpt below (excerpt from "Why Codeine is Dangerous in Some Kids after Tonsillectomy," by David Kroll, February 2013, *Forbes*).

"...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."

As research in genetics continues, the goal is to be able to use information about a person's genetic make-up to find medicines that will be effective with minimal side effects.

After reading this section, answer Question 2 in writing on the worksheet (page 4).

3. To learn about future generations

Prenatal testing: It is possible to use a blood sample from a pregnant person to test the DNA of the fetus they are carrying. This test is known as *non-invasive prenatal testing* (NIPT). NIPT makes information about the genetic make-up of a fetus available earlier in a pregnancy than ever before.

How does NIPT work? All of us have small pieces 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 comes from cells of the placenta, an organ formed by embryonic cells that connects the developing embryo (and later the fetus) to the uterus. By obtaining a blood sample from the arm of the pregnant person, doctors can analyze the cell-free DNA to reveal information about the developing fetus.

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.

Testing embryos created by in vitro fertilization (IVF): Some families know about a genetic condition that runs in their family, such as cystic fibrosis or Huntington's disease. There is a technique that can allow people from these families to avoid passing on this disease to the next generation. This technique, called *preimplantation genetic diagnosis* (PGD), was first performed in 1990.

How does PGD work? First, eggs are fertilized by sperm outside of the body in a laboratory (a process called *in vitro fertilization* or IVF). If the fertilization is successful, this creates an embryo. An embryo can then be implanted into the uterus with the hope this procedure will lead to a pregnancy. For PGD, genetic testing is performed before an embryo is implanted into the uterus. One or more cells are removed from embryos created by IVF. Then, DNA from these cells is tested for various genetic markers, and this information is used to decide which embryos to implant into the uterus.

Cost and access: Access to IVF and PGD is not available to everyone who might want to use these techniques because they are very expensive. In 2020, the average cost of IVF and PGD combined is \$20,000-30,000 US. This procedure is covered by public health care systems in some countries. However, in the U.S., less than one-third of US states require health insurance to include these sorts of treatments. For many people without health insurance, the procedure is out of reach.

After reading this section, answer Question 3 in writing on the worksheet (page 4).

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STUDENT QUESTIONS

Name: _____ Date: _____

1. At the end of Angelina Jolie's op-ed, she states why she decided to go public with her experience. In your words, what were her reasons to share this story with the world?
2. Have you or someone you know ever taken a medicine that didn't seem to work very well? In your own words, how do you think the idea explained above – that genetics might play a role in how some medicines can affect people – could be helpful to people who need to take certain medicines to stay healthy?
3. Write short definitions for the terms "NIPT", "PGD", and explain how they are different from each other.