



Personal Genetics Education Project

Ethical, Legal and Social Issues in Personal Genetics

SNAPSHOT

When New Treatments Come with Big Hopes and a Big Price Tag

Adapted for PBS LearningMedia in partnership with WETA for use with

KEN BURNS PRESENTS
THE GENE
AN INTIMATE HISTORY

2020



HARVARD
MEDICAL SCHOOL

**BLAVATNIK INSTITUTE
GENETICS**

SEPA SCIENCE EDUCATION
PARTNERSHIP AWARD
Supported by the National Institutes of Health

pgEd acknowledges generous support from the Department of Genetics at Harvard Medical School, where we are based. This project was supported by the National Institute of General Medical Sciences, the National Institutes of Health under Award Number R25GM129172. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.



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Big Picture:

- Researchers have developed treatments for people with a rare genetic disorder.
- Families are excited, but the cost prohibits access for people who might benefit.

Rare diseases are not as rare as you might think.

Having what is considered a “rare” genetic disorder is more common than many people might realize. There are 6,800 diseases considered “rare”, although the term is defined slightly differently in different parts of the world. In the United States, the term is applied to diseases that affect fewer than 200,000 people in the US. Together, rare diseases are thought to impact over 300 million people worldwide.

Case Study: Spinal Muscular Atrophy (SMA)

SMA is a rare genetic disease characterized by certain variants in a gene called SMN1. These variants prevent the gene from producing an important protein. When this protein is missing, motor neurons (the nerves that tell muscles to move) function poorly, causing a variety of symptoms. People with some versions of SMA lose or never develop the ability to walk, crawl, or lift their head. Often times, as the disease advances, swallowing and breathing become difficult or impossible. Certain forms of SMA are often fatal before the age of 2.

After years of research, scientific discovery, and activism from families, two SMA treatments have come to market, one in 2016 and one in 2019.

Do Now: Watch [Clip 1](#) from the PBS documentary, *The Gene: An Intimate History*.

In the clip, we meet the Yoders, who have had two children with SMA. When their daughter, Ariel was born, there were no drugs available for SMA. Ariel passed away in April 2014 when she was just 18 months old. Shortly after her death, the Yoders had a son named Jase, who also has SMA. In the short amount of time between the births of these two children, a drug for SMA had been developed and was being tested in a clinical trial. Jase was enrolled in that trial and got his first dose of the medication when he was just 18 days old. He continues to get injections of this medication into his spinal fluid every few months, and he is seeing incredible benefits. The medication Jase is taking, Spinraza (nusinersen), has been approved by the US government for patients of all ages. It is also available in certain other countries around the world. This medication costs approximately \$750,000 for the first year, plus \$375,000 for every year after.

In 2019, the US government became the first, and only, country to approve another promising drug to treat SMA called Zolgensma. Because it is only approved in the US, only people in the US have the potential to access it (for example, through health insurance programs or a research study). This medication appears to halt the onset of the disease if given to babies before the age of 2, and before they develop any symptoms of SMA. This means that, for the medicine to be most effective, a baby needs to have a genetic diagnosis even before they show any symptoms. Zolgensma is the most expensive drug in the world, costing \$2.1 million dollars. Unlike Spinraza, it is a one-dose treatment that is administered intravenously and is only approved for babies under the age of 2. Insurance coverage of the drug varies, and some insurers only pay for it for babies under 6 months old. Patients, advocacy organizations, researchers, insurers, and the drug company producing the medicine all recognize the need to find a way to make this drug more accessible to the people who might benefit. But how?

Do Now: Watch [Clip 2](#) from the PBS documentary, *The Gene: An Intimate History*.

Read the following article and then answer the question below: "[A Lottery Offers Like No Other Offers Up A Cutting-Edge Medicine – With Lives on The Line](#)" by Andrew Joseph and Ed Silverman, February 2020, *STAT News*.

Question: In the article, different people express their views (the pressures, worries, and opportunities) about how to handle the costs and limited availability of SMA drugs, like Zolgensma. After reading the article, summarize the views of three different stakeholders (e.g., a parent, a bioethicist, and a drug company representative). You may select one person from each of the 3 groups below.

1. A parent of a child hoping for access to the SMA drug (parents quoted include Jamie Clarkson, Laura Silva, and Maura Blair)
2. Bioethicists (Holly Fernandez Lynch and Genevieve Kanter)
3. Drug company representatives (Dave Lennon)