



# Personal Genetics Education Project

## Ethical, Legal and Social Issues in Personal Genetics

### SNAPSHOT

## Identifying & Understanding Rare Genetic Conditions: Meet Tess Bigelow

Adapted for PBS LearningMedia in partnership with WETA for use with

KEN BURNS PRESENTS  
**THE GENE**  
AN INTIMATE HISTORY

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## SNAPSHOT

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### Big Picture:

- Why might families affected by a rare genetic disorder look to genetic testing and advances in genetic research?
- What are some of the challenges that these families face?

Watch a video clip from *The Gene: An Intimate History*.

**Do Now:** Watch [this clip](#) from the PBS documentary, *The Gene: An Intimate History*, to meet Luke Rosen. Luke's daughter, Susannah, has a rare disease caused by a change in her KIF1A gene. We follow Luke at a conference, where he hopes to attract the attention of researchers and learn about recent discoveries in conditions related to Susannah's. As we see in the clip, parents of children with rare diseases often find themselves in the role of advocate as well as caregiver. In this lesson, we will learn about the Bigelow family who also has a child, Tess, with a rare genetic disease.

Why are new genetic tools providing hope for children and families affected by a rare and undiagnosed condition?

One major use of genetic testing is to determine whether an illness is genetic, with the hope of identifying a cure or treatment plan. For some people, genetic analysis can reveal a long-sought-after answer when visits to family doctors and then specialists fail to produce a diagnosis. This long and frustrating quest for an answer is sometimes called a "diagnostic odyssey". When a family has a child with a rare genetic disorder, how can they seek out and connect with others to share medical resources and

emotional support? The Bigelows' story, like the Rosens', is an example of how families can take action, even when faced with a medical condition that does not yet have an established treatment – or in this case, even a name.

## Learn about Tess's story.

As a young girl, Tess Bigelow had a number of complex health issues and developmental delays, and so doctors looked in her DNA to try to find out why. The analysis found 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 to search for other children with similar health issues also connected to a change in the USP7 gene. They did not know if any other person in the world had the same condition as Tess. The following day, someone saw Bo's post and connected him with a genetic researcher studying the USP7 gene. Bo learned from the researcher that there were seven other children known to have the same condition as Tess.

**WANTED**  
DELETION OR MUTATION IN THE USP7 GENE ON CHROMOSOME 16

|                                       |  |                                     |   |
|---------------------------------------|--|-------------------------------------|---|
| AMANDA, 16 YEARS OLD - USA - MICHIGAN | ZOE, 4 YEARS OLD - FRANCE - MICHIGAN   | ELLA, 16 YEARS OLD - USA - MICHIGAN | ROSE, 11 YEARS OLD - USA - MICHIGAN     |
| EMILIE, 1 YEARS OLD - USA - MICHIGAN  | CAROL, 4 YEARS OLD - FRANCE - MICHIGAN | MEL, 7 YEARS OLD - USA - MICHIGAN   | EMILY, 11 YEARS OLD - CANADA - MICHIGAN |
| ELLA, 1 YEARS OLD - USA - MICHIGAN    | 8 YEARS OLD In 2015<br>Deletion        | ROSE, 4 YEARS OLD - USA - MICHIGAN  | ...                                     |

In order to help Tess, Ella, Rose, Zoe and other patients find a cure, the foundation for USP7 wants to find other cases with the same pathology (mutation or deletion in the USP7 gene on chromosome 16 (22 cases known in the world). If you know of similar cases, please contact us by mail : [admin@usp7.org](mailto:admin@usp7.org) or through our USP7 Facebook Group. USP7 foundation's aims are to support families affected by such kind of rare diseases and to advance research.

**HELP US FIND OTHER CASES !**

**USP7**  
FOUNDATION FOR USP7 RELATED DISEASES  
11 Ironworkers Lane, Falmouth, ME 04105, USA  
[admin@usp7.org](mailto:admin@usp7.org)

Courtesy of Bo Bigelow, [usp7.org](http://usp7.org)

The Bigelow family have become activists. They are champions for Tess and her medical journey, and they are seeking to find and connect families all over the world who are affected by this same genetic condition. The Bigelow family launched a foundation and created a community of other affected families, hoping one day to find enough patients to populate a research study. Like many families living with a rare disease, the Bigelows dream of research advances and the development of medical treatments.

**Watch this video, as the Bigelow family tells their story:** [Tess Is Not Alone - A USP7 story.](#)

## Answer the following questions.

Try and put yourself in the shoes of Tess's family when they learned for the first time that there were other kids who had the same genetic condition. How do you think you'd feel? What would be going through your mind? In *The Gene*, we meet the Rosen family which also has a child with a rare genetic disease. What similarities do you see between the Bigelows' and the Rosens' stories?