

# Silencing chromosome 21

December 4 2013, by Angela Herring

---



Cell biologist Jeanne Lawrence discussed her work on Down syndrome at the seventh installment of the Profiles in Innovation Presidential Speaker Series. Credit: Brooks Canaday.

Cell biologist Jeanne Lawrence's revolutionary recent discovery of a gene that effectively turns off the chromosome responsible for Down syndrome set the scientific world abuzz. On Monday night at Northeastern, she described how her finding was just as surprising as her journey to becoming a scientist.

The day Lawrence accepted her first research job, she cried. The human genetics counselor had received a fellowship to train in science but she didn't want to be a scientist. She wanted to work with people and make a difference, she told President Joseph E. Aoun, who hosted the seventh installment of the Profiles in Innovation Presidential Speakers Series in the Raytheon Amphitheater.

But after a few months of staring through the microscope, something happened: "I fell in love with chromosomes," said Lawrence, whose deep interest in this work inspired her to earn a doctorate in [developmental biology](#) from Brown University. Better yet, she didn't have to give up on her dream of helping people. In 2007, Lawrence, now professor and chair in the Department of Cell and Developmental Biology at the University of Massachusetts Medical School, received a Eureka grant from the National Institutes of Health to study Down syndrome, which she described as one of the least funded disorders in the world.

Launched in 2011, the Profiles in Innovation Presidential Speaker Series was designed to bring the world's most creative minds to campus for conversations on innovation and entrepreneurship. In keeping with that focus, Aoun described Lawrence as an innovator and credited her determination to tackle this research despite its many risks. "You were a taking a high risk, high reward, and high failure possibility, but you kept going," he said.

Affecting 400,000 people in the U.S. and 6 million people worldwide, the disorder—which is caused by an extra copy of chromosome 21—is a leading cause of cognitive and physical impairment and has been associated with early onset Alzheimer's disease, [congestive heart failure](#), and leukemia.

But Down syndrome research is nowhere near commonplace, and Lawrence suspects one reason for that is the mindset among the research

community. Since it's a chromosomal abnormality, she explained, researchers thought it must be completely intractable. But years of working with the chromosomes that determine sex had given Lawrence a different idea.

Every female has an extra X chromosome, she explained, which means that a natural mechanism for silencing extra [chromosomes](#) already exists. She wondered if that mechanism could be generalizable to chromosome 21. "There are a number of reasons why this won't work," she said. "But no one of them was an absolute."

The gene responsible for silencing the X chromosome, called XIST, is extremely large. No one had ever transplanted such a big sequence into the genome, she said. And even if she could get it from the X chromosome to chromosome 21, there was no guarantee it would behave the same way in its new home.

But Lawrence's high-risk project paid off with high reward: In July of this year her team published the first study to successfully silence the extra copy of chromosome 21 using XIST.

Her work could lead to much greater understanding of Down syndrome's symptoms and treatment methods, but Lawrence was quick to point out that she does not have a cure. "I don't envision that that's going to be possible," she said. "What we're thinking about is that list of conditions and medical challenges that affect an individual with Down syndrome."

While that prospect of helping people compelled Lawrence's journey, it began with a love for the science. "If you love doing what you're doing, that's a really big reason to do it," she said. "The process of working on it is your reward."

Following Lawrence's talk, she fielded questions from the audience

members and via social media on topics ranging from how silencing [chromosome 21](#) would affect cognition in people with Down syndrome to how to prepare for failure in high-risk research. One audience member shared that her sister has Down syndrome and while she is hopeful about the positive health impacts a potential chromosomal therapy could have, she "raises an eyebrow" at the prospect of turning off Down syndrome entirely. In response, Lawrence reiterated that the findings would not lead to treatments that could change an individual's fundamental nature.

Provided by Northeastern University

Citation: Silencing chromosome 21 (2013, December 4) retrieved 19 April 2024 from <https://medicalxpress.com/news/2013-12-silencing-chromosome.html>

<p>This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.</p>
--