Establishing a Rare Disease Advisory Council—Supporting 65,000 Vermonters

A presentation to the House Committee on Human Services
Wednesday, 16 April 2025, at 10:00 AM, Room 46, VT State House
A personal statement by Leslie Kanat

My name is Leslie Kanat and I live in Jericho VT with my wife, Lanie, and our two children. I started teaching geology on the Johnson State Campus in 1989. In 2020 I resigned from my faculty position to devote more time with the family. Presently, I serve as Dean of the School of Science, Technology, Engineering, and Mathematics at Vermont State University. I am here today to ask for your support to establish a Rare Disease Advisory Council, but first, I want to share a story with you.

Our son, Ryan, has Fragile X Syndrome (FXS). He was diagnosed when he was four years old, yet we knew something was very wrong soon after birth. Our daughter, Marin, was one year old at the time and a blood test ruled out FXS. FXS is a family of genetic conditions caused by changes in one gene, FMR1 (Fragile Mental Retardation 1), on the X chromosome. It is the most common inherited form of cognitive impairment and displays a broad spectrum of emotional and behavioral involvement, including autism, anxiety, hyperactivity, avoidance of eye contact, and seizures.

FXS is a medical condition. When the gene does not function, the protein (FMRP) is not made, and the result is FXS. FXS includes i) Fragile X Tremor Ataxia Syndrome, an age-related condition like Parkinsons and with associated with balance issues, and ii) Fragile X Primary Ovarian Insufficiency that results in infertility, early menopause, and other ovarian problems. The gene was discovered in 1991, and the inheritance patterns are now understood. There is no cure for Fragile X Syndrome.

My wife and I are caregivers and legal guardians for our 23-year-old son. He does not communicate clearly, he lacks numerical literacy, he cannot advocate for himself, and he cannot take care of himself. He acts on impulse, not thoughtful decision making, so he is never left alone. We cannot find appropriate medical support or services in Vermont, we need help finding personal care assistants, and we need to find a safe place for him to live. FXS is a rare disease, yet my situation is not unique. Many Vermonters with different rare diseases have similar problems.

The US Food and Drug Administration defines a rare disease as a condition that affects fewer than 200,000 people in the country at any given time. The US Food and Drug Administration estimates that more than 30 million people in the US have a rare disease, which is roughly 1 in 10 people, or approximately 65,000 Vermonters.

I ask this Committee to help us establish a Rare Disease Advisory Council in Vermont. Representatives Donahue of Northfield, Noyes of Wolcott, and Stone of Burlington introduced H.46 that uses language drafted by me and a coalition of Vermont-based advocates organized by the National Organization of Rare Diseases, to form a Rare Disease Advisory Council.

Establishing a Rare Disease Advisory Council will enable Vermont to strategically identify and address barriers that prevent Vermonters with rare diseases from obtaining proper treatment, finding care, and learning about where they can go for support. A Rare Disease Advisory Council is an advisory body providing a platform for the rare community to have a stronger voice in state government. I ask you to advance H.46 through this Committee so we can pass it in the House before the session break. H.46 ensures that patients, caregivers, healthcare providers, researchers, and policy experts can help shape better care and support for those with rare diseases. Thank you for your time today and for your service to support all Vermonters.

Fragile X Syndrome is a family of genetic conditions caused by changes in one gene. It is the most common inherited form of cognitive impairment and displays a broad spectrum of emotional and behavioral involvement including autism and behavioral challenges, anxiety, hyperactivity, avoidance of eye contact, and seizures.

New situations may cause a person with Fragile X to become anxious or afraid.

Thank you for your understanding.

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Fragile X Syndrome

One in 130 women carry the Fragile X gene;
1:6000 females and 1:3000 males are affected.
Older male carriers exhibit tremor ataxia syndrome;
female carriers exhibit primary ovarian insufficiency.

fragilex.org
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fraxa.org

VT Fragile X

vermont@fragilex.org

Katie Couric

Lane, Marin, Les, and Ryan Kanat