

H.46 – RARE DISEASE ADVISORY COUNCIL

Committee Packet Submitted by:

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Committee: Senate Health & Welfare

Purpose

To provide testimony and supporting documents regarding H.46, an act establishing a Rare Disease Advisory Council in Vermont.

1. Written Testimony – Danielle Jankowski
2. Zamara’s Story (Lived Experience Summary)
3. Beckwith-Wiedemann Syndrome (BWS) Overview
4. System Gaps in Vermont’s Rare Disease Care
5. Why H.46 Matters
6. Rare Disease Data & National Context

Statement of Intent

This packet is submitted to support the passage of H.46 and to ensure that Vermont establishes a Rare Disease Advisory Council capable of improving diagnosis, care coordination, provider education, and outcomes for children and families affected by rare diseases. The order of documents reflects the progression from personal testimony, to lived experience, to clinical context, and finally to the systemic evidence that necessitates legislative action.

1. WRITTEN TESTIMONY – DANIELLE JANKOWSKI

Testimony of:

Danielle Jankowski, Pittsford, Vermont

Certified Clinical Medical Assistant & Pediatric Cancer Advocate

Regarding: H.46 – Rare Disease Advisory Council

Good morning, members of the Committee. My name is Danielle Jankowski and I live in Pittsford VT, and I am a Pediatric Cancer Advocate and a Certified Clinical Medical Assistant.

I want to tell you why I am in this field. I didn't just stumble into medicine. I fought my way into it. I became a CCMA to get my foot in the door because I have a very specific mission: I am working toward becoming a Pediatric Oncology Nurse. And I am doing it because of my daughter, Zamara.

I became a medical professional because of what this system did to Zamara. In her journey, I saw firsthand how the system fails when it is confronted with the "rare" or the "complex." I saw the silence that happens when doctors don't have answers. I saw a mother myself being ignored while my child suffered.

I decided then that I would never let that happen to another child if I could help it. I am training to be the voice Zamara needed the voice that I know another child in Vermont will need when they are facing a terrifying diagnosis. But as much as I can do as one nurse or one assistant, I cannot fix a broken system alone. That is why we need H.46.

My fight isn't just in the past; it is my everyday reality. Two of my children live with Beckwith-Wiedemann Syndrome (BWS). Because of this rare overgrowth disorder, my children live with a constant, looming risk of childhood cancer.

Our lives are dictated by "the scan." Every three months, we are in a clinic for ultrasounds, Chest X-rays, Urin Cultures and bloodwork to check for tumors. As someone who is studying for the nursing field, I understand the clinical high-stakes of BWS. But as a mother, I see the gaps in Vermont's care. We are often the ones teaching the providers about the protocols. We are the ones coordinating between specialists who don't talk to each other. If a future oncology nurse is struggling to navigate this system, imagine the parents who have no medical background at all.

H.46 is the bridge between a parent's intuition and a child's survival. A Rare Disease Advisory Council ensures that the lived experience of families like mine is finally used to build a better roadmap for Vermont.

I have done the work. I have changed my career and my life to be the advocate my kids and Zamara deserved. I am asking you to do your part now. Pass H.46 so that when a family hears a rare diagnosis, they aren't met with a wall of silence, but with a system that is ready to listen.

Please pass H.46. Let's make sure that Zamara's name stands for a Vermont that finally learned how to fight for its rarest children.

Submitted for the record.

2. ZAMARA'S STORY (LIVED EXPERIENCE SUMMARY)

Zamara's Story (Neuroblastoma Cancer & misdiagnosed)

Zamara was a bright, beautiful 2 year old child whose symptoms were dismissed and misunderstood. When she needed answers, the system fell silent. When she needed urgency, the system hesitated. When she needed coordinated care, the system fractured.

As her mother, I watched her suffer while my concerns were minimized. As a future nurse, I now understand how preventable some of those failures were. Her journey is the reason I entered medicine. Her name is the reason I am here today.

Zamara's story is not an isolated tragedy it is a warning. And it is a call to action.

3. BECKWITH-WIEDEMANN SYNDROME (BWS) OVERVIEW

Beckwith-Wiedemann Syndrome (BWS): Key Information

What is BWS?

A rare genetic overgrowth disorder affecting approximately 1 in 10,500 births. Children with BWS have a significantly increased risk of developing embryonal tumors, including Wilms tumor, hepatoblastoma, and neuroblastoma.

Common Clinical Features:

- Organ enlargement
- Limb or body asymmetry
- Abdominal wall defects
- Neonatal hypoglycemia
- Increased childhood cancer risk

Standard Cancer Surveillance Protocol:

- Abdominal ultrasound every 3 months from birth to age 8
- AFP blood test every 3 months from birth to age 4
- Chest X-ray (periodic, based on risk and provider guidance)
- Urine catecholamine testing (VMA/HVA) to screen for neuroblastoma
- Immediate evaluation for any abdominal mass, swelling, or unexplained symptoms

Why Surveillance Matters:

Early detection dramatically improves outcomes. Delayed or inconsistent surveillance increases the risk of late-stage cancer, especially for fast-moving tumors like neuroblastoma.

Current Challenges in Vermont:

- Providers unfamiliar with BWS protocols
- Families educating clinicians
- Missed or delayed surveillance
- Fragmented care across specialties

- No centralized rare disease resource
- Parents must fight to secure appropriate screenings, even when national guidelines are clear

4. SYSTEM GAPS IN VERMONT'S RARE DISEASE CARE

1. Lack of Provider Awareness

Families routinely encounter clinicians unfamiliar with rare disease protocols. Even with a known diagnosis like BWS, parents must push for appropriate cancer surveillance, including ultrasounds, AFP testing, chest X-rays, and urine catecholamine screening.

2. Parents Forced to Advocate for Basic Care

Despite clear national guidelines, families often have to fight to secure the screenings their children need. This includes my own experience advocating repeatedly for my own children's BWS surveillance in Vermont.

3. No Centralized Expertise

Vermont has no statewide rare disease resource or advisory body to guide providers or support families.

4. Fragmented Care

Specialists often do not communicate. Parents become the care coordinators, navigating oncology, genetics, primary care, and specialty clinics alone.

5. Inconsistent Surveillance

Children with known cancer risks experience delays or missed screenings due to provider unfamiliarity or lack of coordinated protocols.

6. Emotional & Administrative Burden on Families

Parents must research, advocate, and correct medical misunderstandings while managing fear and uncertainty.

7. No Mechanism for Lived Experience Input

Families with rare disease expertise have no formal seat at the table to inform policy or clinical practice.

5. WHY H.46 MATTERS

Why Vermont Needs H.46 – The Rare Disease Advisory Council

What the Council Will Do:

- Bring together clinicians, researchers, families, and state leaders
- Provide guidance on rare disease care
- Identify gaps in Vermont's system
- Recommend improvements to screening, diagnosis, and treatment
- Ensure lived experience informs policy

Why It Matters:

- Rare disease families currently navigate alone
- Providers lack centralized guidance
- Children face delayed diagnoses and inconsistent care
- Vermont has no rare disease infrastructure

How It Helps Families:

- Creates a clear roadmap for care
- Improves provider education
- Reduces misdiagnosis and delays
- Ensures families' voices shape solutions

How It Helps Vermont:

- Strengthens healthcare quality
- Reduces long-term costs through earlier detection
- Aligns Vermont with national best practices

6. RARE DISEASE DATA & NATIONAL CONTEXT

Key Statistics:

- 1 in 10 Americans lives with a rare disease
- Over 7,000 rare diseases exist
- 80% are genetic
- 50% of rare disease patients are children
- Average diagnostic delay: 5–7 years

States With Rare Disease Advisory Councils:

- Maine
- New Hampshire
- Massachusetts
- New York
- Connecticut
- 25+ states nationwide

Vermont is behind. H.46 closes that gap.