Annual Report 2025
Rare Disease Advisory Council
RSA 126-A (HB237, CH24:1) Laws of 2019
March 27, 2026

Committee Membership:

Mary Beth Dinulos, MD Chair
Angela Shepard, MD
Rep. Efstathia Booras
Rep. David Nagel, MD
Sen. Senator Innis
Nancy Biederman
Laura Landerman-Garber, PhD
Elizabeth Shannon
Courtney Keane
Elijah Stommel, MD
Richard Lafluer, MD

Council in charge and purpose:

(a) Advise the legislature and department of health and human services on rare diseases in New Hampshire
(b) Coordinate with other states' rare disease advisory bodies, community-based organizations, and other public and private organizations for the purpose of ensuring greater cooperation between state and federal agencies may include, but are not limited to, the National Institutes of Health, and the United States Food and Drug Administration.
(c) Explore existing data on rare diseases in New Hampshire collected by the department of health and human services
(d) Encourage public health awareness regarding rare diseases in New Hampshire

In 2025 the NH Rare Disease Advisory Council (RDAC) held meetings in a hybrid fashion every month for one hour. The meeting discussions focused on legislative updates, newborn screening (blood spot), sudden death in the young committee, telehealth, and community outreach and education. The committee welcomed the public to attend and share their experience/story of a rare disease.

Relevant to our charges above, the Council accomplished the following:

(a) Advise the legislature and department of health and human services on rare diseases in New Hampshire

The committee discussed relevant legislation pertaining to patients with rare diseases and their families. Several of the pertinent bills that were followed include:

- Recognizing the geographic challenges and limited access to specialized care, the Council prioritized expanding telehealth services. It supported 2025 HB 72, extending the commission to study telehealth services, and 2025 SB 252, establishing criteria for providing certain medical care through telemedicine. Both bills passed and were signed into law.

- The Council also addressed the impact of Amyotrophic Lateral Sclerosis (ALS) by supporting 2025 HB 576, which aimed to track ALS diagnoses in the state. Although the bill was found inexpedient to legislate, the Council brought the issue to the HHS Oversight Committee to explore more cost-effective ways to collect this data.
- To improve access to a broader range of therapies, the Council supported 2025 HB 241. The bill was retained for further study, amended, and ultimately gained broad bipartisan support. It is scheduled to be heard again in 2026.
- To preserve institutional knowledge and expertise, the Council supported an amendment to 2025 HB 118 eliminating term limits for RDAC membership. This measure passed and was signed into law, allowing continued service from experienced members while still welcoming new participants.
- The Council identified a critical gap in New Hampshire's lack of a step therapy appeals process. In response, it collaborated with the Susan G. Komen Foundation and other nonprofit partners to develop 2026 HB 1638, which would create a medically necessary bypass mechanism for step therapy protocols.
- Additional advocacy efforts included support for 2025 HB 677, requiring epinephrine auto-injectors (EpiPens) in all schools to address sudden, life-threatening allergic reactions and reduce the burden on families who may struggle to provide multiple devices. Testimony highlighted the risks associated with children transporting EpiPens between home and school.
- The Council also supported 2025 HB 701, the "Right to Try" bill, advocating for individuals with life-threatening rare conditions to access potentially life-extending treatments not yet approved by the FDA. Testimony was provided in person, and members attended the bill signing.

(b) Coordinator with other states' rare disease advisory bodies, community-based organizations, and other public and private organizations for the purpose of ensuring greater cooperation between state and federal agencies may include, but are not limited to, the National Institutes of Health, and the United States Food and Drug Administration.

The RDAC explored community-based organizations and other public and provide organizations to learn how to improve NH RDAC.

The NH RDAC continues to partner with the National Organization of Rare Disorders (NORD) as well as other state RDACs to network and discuss topics important to the rare disease community such as PDABs and drug pricing, Medicaid coverage, and the role of the FDA in rare disease drug development.

The RDAC has continued to develop relationships with other organizations including the NH Council on Developmental Disabilities, the NH Rare Disorders Association, the NH Rare Action Network, Rare New England and the Sudden Death in the Young Committee with participation in council meetings and communication. Dr. Mary Beth Dinulos, current Chair of the NH RDAC, was an invited speaker at the Rare New England annual conference at Southern NH University in November 2025. Her talk entitled "Getting Involved: Navigating Your State's RDAC for Meaningful Change" educated members of the rare disease community on how to get

involved in rare disease advocacy. Dr. Dinulos also had a platform presentation at the March 2025 American College of Medical Genetics and Genomics annual meeting in Los Angeles, CA. She discussed the importance of the NH RDAC in her work as a medical geneticist caring for patients with rare diseases.

(c) Explore existing data on rare diseases in New Hampshire collected by the department of health and human services

The committee discussed having a database that can be accessed to understand NH residents who have rare diseases and how best to support the needs of individuals and families.

(d) Encourage public health awareness regarding rare diseases in New Hampshire

This past year the NH RDAC focused a great amount of effort on public health awareness of rare diseases. Below is a list of activities:

February–April 2025

- Attended meetings during Rare Disease Advocacy Week with the Everylife Foundation and met with state representatives to advocate for the "Kids' Access to Care Act."
- Continued advocacy efforts by submitting letters to newspapers, including the *Concord Monitor*, *The Manchester Union Leader*, *Laconia Daily Sun*, and *Conway Daily Sun*.
- Broadcast a public service announcement throughout February on WFEA (Manchester Radio Group) to raise awareness about rare diseases and promote Rare Disease Day.
- Participated in WFEA segments, including *Eye on New Hampshire with Marga* (Laura and Nancy discussed challenges facing the rare disease community) and *The WFEA Morning Update with Jeff Chidester* (Nancy shared statistics and impacts of rare diseases in New Hampshire).
- Hosted a Rare Disease Day event, including lighting the Memorial Bridge in Portsmouth in purple (sponsored by New England Connection for PKU and Allied Disorders, Inc.) and tabling at the State House Cafeteria to share information and distribute cookies and zebra ribbons.

May–July 2025

- Continued local advocacy efforts, including submitting testimony on state House bills related to medication coverage.

August 2025

- Participated in the "Rare Across America" Advocacy Week with NORD and the EverIylife Foundation, including meetings with federal representatives such as Congressmember Chris Pappas.

<u>Specific issues addressed by members:</u>

Sudden Death in the Young Committee

Dr. Dinulos is section chief of Genetics and Child Development at DHMC and a member of the NH Sudden Death in the Young (SDY) committee. This committee reviews all sudden deaths in

children and infants (SUID - Sudden Unexplained Infant Death) in the state of New Hampshire. Criteria include:

a) All cases must be residents of NH
b) Children are less than 20 years old and have sudden and unexpected death
c) Includes drownings in children older than age 5 years and drivers in motor vehicle accidents

At the end of 2025, New Hampshire transitioned its molecular genetic testing services to Dartmouth Health. This shift is a significant enhancement in the state's approach to supporting families who have lost a child suddenly and unexpectedly. Through this partnership, the SUID/SDY program, in collaboration with the Office of the Chief Medical Examiner, has the support and access of specialized genetic counseling services. This ensures that families are presented with the test results, but also have an expert to guide and help understand the results. Importantly, genetic counselors can also assess whether additional testing may be appropriate for immediate family members, helping to identify inherited risks and inform preventative care. This more comprehensive approach strengthens both clinical follow-up and long-term public health outcomes by improving early detection, risk assessment, and access to appropriate medical interventions.

Dr. Dinulos continues to advocate for molecular genetic testing in these children with unexpected death. She works closely with the SDY case registry manager, the director of the NH SDY committee and the medical examiners in order to maintain funding for molecular genetic testing in these children.

NH Newborn Screening Program

The Newborn Screening Program Advisory Committee reviewed, discussed, and voted in 2025 on the addition of three disorders: GAMT, MPS II, and Krabbe disease. During the advisory committee meetings, members heard from families, representatives from the University of Massachusetts Chan Medical School (where the testing is done), and a metabolic specialist. This information allowed the committee to determine that each disorder is well defined with a known incidence; associated with significant morbidity and/or mortality; detectable through a screening test that is ethical, safe, accurate, and cost-effective; and whether effective treatment exist, especially if early treatment, prior to symptom onset, leads to better health outcomes than later intervention. Based on this information, the committee voted in October 2025 to approve the addition of all three disorders. The disorders will be phased in over the course of 2026/2027 to allow sufficient time for both the program and the laboratory to establish all necessary programmatic and clinical protocols. With the addition of these 3 disorders, NH will screen 39 disorders in total, all of which are on the recommended uniform screening panel (RUSP).

Courtney Keane, DHHS representative on the NH RDAC, is the Bureau Chief for Family Health and Nutrition, which oversees the NH newborn screening program. She keeps the Council

informed regarding the status of newborn screening in the state, and alerts the Council to any upcoming legislation regarding NBS.