Annual Report
Rare Disease Advisory Council
RSA 126-A (HB237, CH24:1) Laws of 2019
March 28, 2025

Committee Membership:

Mary Beth Dinulos, MD Chair
Angela Shepard, MD
Rep. Frances Nutter- Upham
Rep. David Nagel, MD
Sen. Cindy Rosenwald
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) 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.
(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 2024 the Rare Disease Advisory Council held meetings in a hybrid fashion every other 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 included:

- SB354 concerning pharmacy benefits, cost sharing requirements and/or out of pocket maximums under health insurance policies as they pertain to Rare Disease treatments. This bill died in chamber.

- NH HB 355 relative to newborn screening for cytomegalovirus, which passed and was signed by the Governor on July 26, 2024.
- NH SB302 relative to physician licensures, which passed and was signed by the Governor on 07/03/2024.

(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.

Two members of the NH RDAC attended the National Organization of Rare Disorders (NORD) 2024 RDAC Members Meeting and NORD Breakthrough Summit held in Washington, DC in October 2024. This gathering brought together representatives from 13 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, and the Sudden Death in the Young Committee with participation in council meetings and communication.

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

The committee discussed having a data base 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 2024

- Attended Rare Disease Events by RDLA Everylife which included online meetings with Senator Maggie Hassan, Senator Jean Shaheen, Congresswoman Annie Kuster and Congressman Chris Pappas
- Attended Rare Disease Day conferences with NORD and NIH
- Hosted Rare Disease Awareness event at NH Capitol with Dr. Laura Landerman and Nancy Biederman
- Guest on WMUR for Rare Disease Day

- Patient-Community Outreach at Kingswood Regional High School - student meeting with Governor Chris Sununu
- Set up Rare Disease Day Awareness display at local library and Infusion Centers

March -April 2024

- Wrote letters to local NH newspapers including The Union Leader concerning public awareness for The Accelerating Kids Access to Care Act HR 4758/S. 2372

May -June -July 2024

- Continued Patient Outreach by connecting with several NH parents and caretakers of Rare Disease patients
- Met with one of the original members of the NH Newborn Screening Council who shared his story of his daughter's medical journey

Specific issues addressed by members:

ALS Database

Dr. Stommel has continued to express interest to have a reportable NH ALS Registry similar to MA, ME, VT and CA.

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

Dr. Dinulos continues to advocate for genetic testing in these children with unexpected death, including comprehensive cardiac panels and comprehensive epilepsy panels. 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 genetic testing in these children.

NH Newborn Screening Program

There are many rare neurological disorders that may have treatments available within the next decade. Identifying the genetic basis of these disorders is often useful for treatments. An example of this is the new treatment for spinal muscular atrophy (SMA) in children. NH screens for SMA through the newborn screening program.

Courtney Keane, DHHS representative on the NH RDAC, is the NH NBS Program Manager. She keeps the Council informed regarding the status of newborn screening in the state, and alerts the Council to any upcoming legislation regarding NBS.