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MAURA T. HEALEY
GOVERNOR

KIMBERLEY DRISCOLL
LIEUTENANT GOVERNOR

KATHLEEN E. WALSH
SECRETARY

ROBERT GOLDSTEIN, MD, PhD
COMMISSIONER

Annual Report
Rare Disease Advisory Council

December 2024

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Executive Summary

The Massachusetts Rare Disease Advisory Council (RDAC) is pleased to present its 2024 annual legislative report as required by the Acts of 2020 Chapter 260 (M.G.L. c.111 s.241). The aim of this report is to inform interested parties, including the governor, the legislature, and the Massachusetts Department of Public Health (DPH) about the progress and ongoing needs of those impacted by a rare disease in Massachusetts. It highlights the work of the council and sets the stage for future initiatives and recommendations.

The RDAC was productive in 2024. The council held 25 full council and working group meetings over the course of the year, including participation at Rare Disease Day at the statehouse in February 2024 and at a legislative briefing at the statehouse in March 2024. Through these meetings, the RDAC focused on five areas that impact the lives of people with rare diseases: 1) newborn screening, 2) medical nutrition, 3) legislation/policy, 4) economic burden, and 5) community engagement.

Newborn Screening

The council undertook a review of the Massachusetts Newborn Screening Program (NSP). This program is a critical asset to families in the Commonwealth for the early identification of children with rare diseases and conditions. With a goal of reinforcing the NSP's mission, the RDAC consulted with the chair of the Massachusetts Newborn Screening Program Advisory Committee (NBSAC), the director of New England Newborn Screening Program (NENSP, of which MA is a part) and other DPH officials. The RDAC subsequently developed recommendations intended to strengthen the NSP. The recommendations are divided into six sections.

1. Strengthen operations of the NBSAC.
2. Increase transparency of the NBSAC.
3. Clarify the evaluation process used to evaluate results of NSP pilot studies, to determine the appropriateness of those conditions for inclusion in the Massachusetts mandated disease screening panel.
4. Increase the timeliness of the NBSAC evaluation of diseases included on the federal Recommended Uniform Screening Panel (RUSP).
5. Establish a NBSAC process for individuals to nominate diseases for addition to the Massachusetts newborn screening panel.
6. Improve outreach and education about mandated newborn screening and pilot programs in Massachusetts.

Notably, the RDAC learned that several of these recommendations were already under consideration for implementation by the NBSAC/NENSP.

Medical Nutrition

The council reviewed the state of medical nutrition supports for people with rare diseases in Massachusetts. Through testimony of subject matter experts, the RDAC learned that Massachusetts has an existing statute that provides coverage for medical food and formula for all inborn errors of metabolism and other select conditions requiring medical nutrition but some limitations in coverage exist. The RDAC plans to investigate this topic further in 2025.

Legislation/Policy

The council proposed to develop a working group to better understanding the legislative/policy landscape related to rare diseases. Council members heard testimony on the legislative process and bills related to rare disease under consideration by the legislature. Upon consultation with the Massachusetts Department of Public Health, the RDAC established a five-person policy working group to identify and track legislation and other federal, state (including state agencies), and municipal policies that may impact the rare disease community from a medical, social, economic, or healthcare/service access perspective and to increase awareness of such legislation and policies through education and outreach.

Economic Burden of Rare Disease in Massachusetts

The council initiated a discussion on the economic burden of rare diseases in Massachusetts. The RDAC heard initial testimony from people living with a rare disease that suggests the economic impacts of living with a rare disease are financially debilitating on a personal level and are also a large driver of health care expenditures at the state level. The RDAC plans to develop a proposal to study the economic burden of rare diseases more fully as a focus area for 2025.

Community Engagement

The council developed a plan to improve community engagement. The plan includes proposed changes to the website to make it more user-friendly. The council also explored new ways to engage with the rare disease community. Regional in-person town hall events were discussed as a potential mechanism to increase community awareness. The RDAC will attempt to establish a working group on community engagement in 2025.

Finally, it is important to note that RDAC membership has undergone significant turn over in 2024. Because the council was formed in 2021, the three-year terms of many members ended in 2024. Most of the members with expiring terms decided to return to the council for another three-year term, while a minority have decided not to continue or have resigned. As part of the effort to replace departing members, the council welcomed six new members in 2024 and awaits the appointments of eight proposed members. The council looks forward to having a fully seated 29-member council in 2025.

Letter from the Chair

December 31, 2024

As Chair of the Massachusetts Rare Disease Advisory Council, I am pleased to share this year's accomplishments and the progress we have made toward our mission of supporting individuals and families impacted by rare diseases.

This year, the council focused on the Massachusetts Newborn Screening Program, recognizing its critical role in early diagnosis and intervention for children with congenital rare diseases. Through testimony from parents and patient advocates, healthcare providers, genetic specialists, and the New England Newborn Screening Program (NENSP), we developed a series of recommendations to strengthen newborn screening for rare diseases in the Commonwealth. Among these recommendations were measures to enhance the oversight and effectiveness of the Newborn Screening Program Advisory Committee (NBSAC), and we are pleased to report several of these improvements are already being implemented. These actions are vital steps toward ensuring Massachusetts remains a national leader in newborn screening.

Looking ahead, we are exploring the possibility of conducting a comprehensive study on the economic burden of rare diseases in Massachusetts. Such a study would provide valuable insights into the healthcare, social, and economic challenges faced by rare disease patients and their families. By quantifying these impacts, we aim to build a data-driven case for policies that addresses the unique needs of the rare disease community. We believe understanding the full economic impact of rare diseases will be a powerful tool for advocating for sustainable support and resources.

Throughout the year, our full council met every other month and collaborated closely with the Massachusetts Department of Public Health (DPH) to align our agenda with state health initiatives. This partnership has been instrumental in advancing our shared mission, providing the council with the necessary support and resources to achieve our goals. As always, we are grateful for the dedication and commitment of our council members and partners. Together, we continue to work toward a future where early diagnosis, effective treatment, and comprehensive support are accessible to everyone affected by rare diseases.

I invite you to review this report and join us in our mission to improve the lives of those with rare diseases in Massachusetts.

Sincerely,

Dylan Tierney

Dylan Tierney, MD MPH
Chair, Rare Disease Advisory Council
Associate Medical Director
Bureau of Infectious Disease and Laboratory Sciences
Massachusetts Department of Public Health

Background

A rare disease is one that affects fewer than 200,000 people in the United States, as defined by the 1983 Orphan Drug Act. Over 7,000 identified rare diseases collectively impact millions of people in the United States. Advances in medicine have made it possible to diagnose new rare diseases every year, so the number of rare diseases continues to grow.

Rare diseases encompass a wide range of disorders. These include genetic disorders, rare cancers, autoimmune diseases, and neurologic diseases, among many others. Approximately 80% of rare diseases have a genetic origin, often resulting from a single gene mutation that can be inherited or occur spontaneously. Rare diseases can affect individuals of all ages, but a significant portion of new diagnoses are made in children and young adults. Many rare diseases are chronic, progressive, and life-threatening. They often present early in life, can be disabling, and can affect physical and mental abilities. Rare diseases can have a significant psychosocial impact on patients and their families, including mental health and financial burdens.

Rare diseases, by their nature, create diagnostic challenges. They are often difficult to diagnose due to their rarity and the lack of awareness among healthcare professionals. The time to diagnosis can be several years, involving periods of misdiagnosis and unnecessary treatments. Treatments also can be limited. Due to the small patient populations of a specific disease or disorder, there is often no commercial interest in developing treatments. Advancements in genomics and personalized medicine, however, are beginning to offer new possibilities for treatment options for those diagnosed with a rare disease.

Massachusetts has a robust healthcare infrastructure and is a leader in diagnosing, treating, and researching rare diseases, according to the National Organization for Rare Disorders (NORD). Although Massachusetts has made notable progress in addressing the needs of individuals with rare diseases, significant challenges remain. The council recognizes that collaboration and coordination with rare disease stakeholders, patients, and advocates will be necessary to ensure equitable access to diagnosis, treatment, and support services for all affected by a rare disease.

Statutes and Mandates

The Massachusetts Rare Disease Advisory Council (RDAC) was established to address the unmet needs of the rare disease community as part of Senate Bill 2984, An Act Promoting a Resilient Healthcare System that Puts Patients First. https://malegislature.gov/Laws/GeneralLaws/PartI/TitleXVI/Chapter111/Section241 . The legislature enacted the bill in December 2020, and Governor Charlie Baker signed it into law on January 1, 2021.

To achieve its purpose, the council's legislative charges include:

i. Coordinating the performance of the rare disease advisory council duties with those of other rare disease advisory bodies, community-based organizations, and other public and private organizations with the Commonwealth to ensure greater cooperation regarding the research, diagnosis, and treatment of rare diseases. This coordination may require:
    a. Disseminating the outcomes of the advisory council's research and identifying best practices and policy recommendations
    b. Utilizing common research collection and dissemination procedures

ii. Using existing publicly available records and information to undertake a statistical and qualitative examination of the prevalence and causes of rare diseases to develop a profile of the social and economic burden of rare diseases in the Commonwealth.
iii. Receive and consider reports and testimony from expert individuals, the Department of Public Health, community-based organizations, voluntary health organizations, healthcare providers, and other public and private organizations recognized as having expertise in rare disease care to learn about their contributions to rare disease care and the possibility to improve rare disease care in the Commonwealth.
iv. Develop methods to publicize the profile of the social and economic burden of rare diseases in the Commonwealth to ensure that the public and health care providers are sufficiently informed of the most effective strategies for recognizing and treating rare diseases.
v. Determine the human impact and economic implications of early treatment of rare diseases versus delayed or inappropriate treatment of rare diseases as it pertains to the quality of care, the quality of patient's and family's lives, and the economic burdens, including insurance reimbursements, rehabilitation, hospitalization, and related services, on patients, families, and the Commonwealth.
vi. Evaluate the current system of rare disease treatment and available public resources to develop recommendations to increase rare disease survival rates, improve quality of life, and prevent and control risks of co-morbidities for rare diseases based on the available evidence.
vii. Research and determine the most appropriate method for the Commonwealth to collect rare disease data, including a database of all rare diseases identified in the Commonwealth along with known best practices for the care of said diseases and such additional information concerning these cases as the advisory council deems necessary and appropriate to conduct thorough and complete epidemiological surveys of rare diseases, subject to all applicable privacy laws and protections.
viii. Examine the feasibility of developing rare disease information and patient support networks in the Commonwealth to aid in determining any genetic or environmental contributors to rare diseases.
ix. Develop and maintain a comprehensive rare disease plan for the Commonwealth utilizing any information and materials received or developed by the advisory council pursuant to the law. The plan shall include information specifically directed toward the general public, state and local officials, state agencies, private organizations and associations, businesses, and industries.

The legislative language allows the advisory council to accept and solicit funds, including gifts, donations, grants, bequests, or federal funds, to carry out the above charges.

Prevalence in Massachusetts

The Massachusetts RDAC has estimated roughly 700,000 Massachusetts residents (1 in 10) may live with rare diseases. However, estimating the prevalence of rare diseases in Massachusetts has been challenging due to underreporting, misdiagnosis, and the steadily increasing number of rare diseases being identified. These limitations highlight the need for improved data collection and reporting mechanisms.

Rare Disease Care in Massachusetts
Massachusetts is one of 24 states that is recognized to have a Rare Disease Center of Excellence as determined by the National Organization for Rare Disorders (NORD). Centers of Excellence are located at Boston Children's Hospital (Genetics and Genomics), Massachusetts General Hospital (Genetics and Genomic Medicine), and Brigham and Women's Hospital (Genetics and Genomic Medicine Service). These Centers are dedicated to diagnosing, treating, and researching all rare diseases and are modeled after healthcare and research networks focused on a single rare disease or a small group of rare diseases. The Centers participating in this network are committed to sharing knowledge and best practices to improve rare disease care and advance rare disease research. These collaborations and partnerships will improve care and better the lives of rare disease patients and their families.

Massachusetts has many other resources for treating rare diseases, including research centers and medical institutions. These include:

- The Manton Center for Orphan Disease Research is located at Boston Children's Hospital. Its goal is to develop ways to diagnose, treat, and understand rare genetic conditions.
- Li Weibo Institute for Rare Diseases Research is located at the University of Massachusetts Chan Medical School. This institution aims to develop new treatments for rare diseases by combining the expertise of its faculty with innovative research platforms.
- Children's Rare Disease Collaborative (CRDC) located at Boston Children's Hospital. This group conducts genomic research on rare diseases to help develop precision medicine for children.

Additionally, many licensed clinicians specializing in rare diseases provide care in settings ranging from tertiary care centers to outpatient clinics across the Commonwealth.

RDAC Membership
The RDAC comprises healthcare professionals, researchers, patient advocates, patients, legislators, and representatives from state agencies. This diverse membership was stipulated in the establishing legislation to foster a comprehensive approach to addressing the needs of the rare disease community in Massachusetts.

The council consists of 29 members. The chair is a designee of the Commissioner of Public Health. One is a designee of the Massachusetts Health Policy Commission. The governor appoints 15 members, the House of Representatives appoints six members, and the Senate appoints six members. Council members serve a three-year term. Most of the founding members were appointed in 2021 and most terms ended in 2024. Seventeen members have chosen to remain for a second term, and four have either resigned or moved out of state. For the remainder of positions, new members have either been sworn in or are in the process of review by the respective appointing body.

New appointments in 2024 include Dr. Christelle Achkar, Dr. Olaf Bodamer, Yue Huang, Dr. Heather Gray-Edwards, and Representative Joseph McKenna. Dr. Achkar replaced Dr. David Miller, whose term expired in May 2024. Dr. Bodamer replaced Dr. Andrew Lane, whose term expired in June 2024. Yue Huang replaced Celia Segel, who resigned. Dr. Gray-Edwards

replaced Dr. Michael Green, who passed away in 2023. Representative Joseph McKenna replaced Representative Hannah Kane, who resigned in February 2024.

The following members also resigned in 2024 but have yet to be replaced: Janis Creedon, Lisa Deck, Guadalupe Hayes-Mota, and Michele Rhee. Candidates for replacement membership have been proposed to the respective authorizing bodies and approval is pending.

The council acknowledges and thanks the following members whose terms have ended but who have remained on the council until their replacement is appointed: Dr. Charlotte Boney, Jeff Livingstone, Diane Lucente, and Ann Wessel.

The list of RDAC members as of December 2024 can be found in Appendix A.

Rare Disease Advisory Council Accomplishments in 2024
Actions taken and progress made towards fulfilling the RDAC mission to improve the lives of people living with a rare disease include:

- Twenty-five meetings
  - Six meetings of the Full Council
  - Five meetings of the Steering Committee
  - Fourteen working group meetings
- The formation of a standing subcommittee
  - RDAC Policy Working Group
- Legislative Briefing
- Participation in the Massachusetts Rare Disease Day
- Participation in the National Organization for Rare Disorders (NORD) RDAC Collaboration Conference
- Consultation with state, federal, and other experts on rare diseases

The council's priorities are driven by its legislative mandates. In 2024, the council prioritized its efforts in five areas that impact the lives of people with rare diseases: 1) newborn screening, 2) medical nutrition, 3) legislation/policy, 4) economic burden, and 5) community engagement.

Newborn Screening
Newborn screening provides an early opportunity to detect certain congenital disorders, often before symptoms appear. Babies with these disorders need treatment in early infancy to prevent severe disease later. Testing all babies is important because most babies appear healthy at birth, even babies who have disorders that need treatment.

The newborn screening program in Massachusetts aims to test all newborns for treatable disorders (as mandated by Massachusetts Department of Public Health Regulations 105 CMR 270.000). Newborn screening is a public health service. Massachusetts has two types of newborn screening: required (mandated) and optional. The required newborn screening is mandated for all newborns except those with a religious exemption. Each year, approximately 160 babies are born in Massachusetts with some rare disorder that is found with required newborn screening. Massachusetts first began this testing in 1962.

In Massachusetts, the Department of Public Health (DPH) is responsible for the oversight of the Massachusetts Newborn Screening Program (NSP). DPH contracts with the University of Massachusetts, New England Newborn Screening Program (NENSP) to ensure that all infants born in Massachusetts have access to newborn screening.

The Newborn Screening Advisory Committee (NBSAC) is a body of individuals appointed by the DPH commissioner to guide the Massachusetts NSP. This committee works closely with the commissioner to determine what disorders are screened in the program. There are currently 32 disorders included in the required screening by the Massachusetts Department of Public Health. Newborns are screened for laboratory markers of the following 32 disorders: https://nensp.umassmed.edu/screening-programs/massachusetts/required-disorders.

The Massachusetts DPH may authorize and direct research studies of new screenings tests through pilot studies in the newborn screening program. Participation in testing through pilot studies is optional. In determining whether a pilot study screening test should be adopted for mandated screening, DPH seeks information on one or more of the following questions:

- What is the extent of benefit from newborn screening for these disorders? (Does it save lives? Does it prevent serious life-compromising outcomes? Do the treatments work as expected?)
- How often do these disorders occur in Massachusetts?
- How good are the laboratory tests used to screen for these disorders?

RDAC Recommendations to Improve Newborn Screening in Massachusetts

The Massachusetts newborn screening program is a critical asset to families in the Commonwealth for the early identification of children with rare diseases and conditions and serves as a role model for the nation. With a goal of reinforcing the program's mission, the RDAC consulted with the chair of the NBSAC and the director of NENSP and DPH officials and have provided the Department with a set of considerations and recommendations.

Recommendations

1. Strengthen operations of the NBSAC.
    - Prioritize robust NBSAC membership as outlined in regulation 105 CMR 27.005(B) and ensure that current members maintain active participation.
    - Establish a NBSAC membership position for a member of the RDAC appointed by the Commissioner.
    - Establish a minimum frequency for NBSAC meetings, as outlined in Acts 2024, Chapter 186 (An Act Promoting Access to Midwifery Care and Out-of-Hospital Birth Options) Section 18.
    - Develop and make public a NBSAC standard operating procedures (SOP) that includes but is not limited to:
        - Purpose,
        - Scope,

    - Membership (roles and responsibilities, nomination process for appointing, maintaining, and replacing membership, term - including the number of terms allowed),
    - Meeting cadence including how often and when the committee meets.
- Explore increased administrative support from DPH for management of NBSAC.
- Develop an annual process to investigate emerging newborn screening technologies and results from NENSP pilot studies and report to the Commissioner on how those technologies and screening opportunities might impact the Massachusetts newborn screening program.
- Develop an SOP that details the referral process for confirmatory testing and care of a newborn that screens positive through the Massachusetts newborn screening program.

2. Increase transparency of the NBSAC.
    - Develop a regular NBSAC meeting schedule and make it public.
    - Post all NBSAC meeting agendas and minutes for the past five years.
    - Publicly list current NBSAC members and their affiliations.

3. Clarify the evaluation process used to evaluate results of NSP pilot studies, to determine the appropriateness of those conditions for inclusion in the Massachusetts mandated disease screening panel.
    - Review pilot studies after two years and make a recommendation to the Commissioner of DPH regarding whether to move the studied condition to the mandated newborn screening panel.
    - Seek public input from parents and medical specialists treating children with the condition on why the condition should or should not be recommended to the mandated newborn screening panel.
    - Provide written justification regarding the conclusion of pilot studies, either to discontinue screening for a pilot condition or to move such screening to the mandatory panel.

4. Increase the timeliness of the NBSAC evaluation of diseases on the RUSP.
    - If the federal Department of Health and Human Services has approved a new disease for inclusion on the Recommended Uniform Screening Panel (RUSP), that disease should be considered for adoption by MA NBS within 90 days of the condition's inclusion on the RUSP.
    - Adopt a policy of requiring expert consultation on any RUSP condition being considered for adoption as a mandate condition for screening on the MA Newborn Screening Panel.
    - Provide a written explanation for NBSAC decisions to accept or reject conditions on the RUSP including reasons for acceptance/rejection.
    - Consider revising criteria in regulation 105 CMR 270.004 for diseases and disorders to be tested as a part of the Massachusetts mandated newborn screening panel to include conditions:

- in which early diagnosis may enable the use of therapeutics that have benefits outside of the newborn infant period.
- in which standard of care treatment is beneficial but may only be available on a limited basis, given that individuals with rare conditions may take extraordinary measures to access these treatments (e.g., travel out of state for treatment).
- in which the benefits – broadly conceived to include the advantages of early diagnosis and its impacts on the care of the child beyond treatment – outweigh the risks and burdens of screening.

5. Establish an NBSAC process for individuals to nominate diseases for addition to the Massachusetts newborn screening panel.
   - Implement a standard operating procedure for the process of receiving nominations for diseases to be consider for addition to the Massachusetts newborn screening panel, including details on:
     - minimum application requirements that the sponsoring group must meet,
     - availability or resources in support of sponsors who would like to submit an application that meets minimum requirements,
     - review and delivery of decision to accept or reject the nominated disease within 90 days of nomination (or emergently if treatment has become available), and
     - ensuring that a written explanation of the decision to accept or reject the nominated disease will be made publicly available.

6. Improve outreach and education about Massachusetts mandated newborn screening and pilot programs.
   - Re-evaluate the educational materials for NBS regarding the differences between the mandatory and pilot screening programs.
   - Develop supplemental educational materials to explain the newborn screening program to Massachusetts families more fully, including simplification of the parental brochure according to health literacy, American Disabilities Act, and plain language principles.

Notably, the RDAC learned that several of these recommendations were already being considered for implementation by the NBSAC/NSP as of November 2024.

Medical Nutrition

Under the Federal Food, Drug, and Cosmetic Act, medical nutrition is defined as "a food which is formulated to be consumed or administered enterally under the supervision of a physician and which is intended for the specific dietary management of a disease or condition for which distinctive nutritional requirements, based on recognized scientific principles, are established by medical evaluation."[1] Many rare disorders require medical nutrition to prevent serious disability,

1. [1]Federal Food, Drug, and Cosmetic Act, Title 21 U.S.C. § 360ee (2015). https://www.govinfo.gov/content/pkg/USCODE-2015-title21/html/USCODE-2015-title21-chap9-subchapV-partB-sec360ee.htm

allow for normal growth in children and adults, or provide adequate caloric intake. Medical nutrition may be the only viable treatment option available for some rare diseases.

Through testimony of subject matter experts, the RDAC learned that Massachusetts has an existing statute that provides coverage for medical food and formula for all inborn errors of metabolism and other select conditions requiring medical nutrition, but some limitations in coverage exist. This program is offered by the Massachusetts DPH and provides access to low-protein foods and formulas for those who qualify. The program offers up to $5,000 in food products to eligible individuals. To participate in the program, the applicant must be enrolled in an eligible MassHealth plan or have other insurance circumstances that meet the program requirements. Although this program only helps a limited number of individuals, it was important to note that it exists.

The council will continue to discuss this topic in 2025 and, if appropriate, will make recommendations on improving access to medical nutrition care for those with rare diseases in Massachusetts.

Development of a Policy Working Group Subcommittee

The council has formed a standing working group to identify and track legislation that may impact the rare disease community. The working group will be chaired by council members Jenn McNary and Glenda Thomas. The working group will consist of five members, and it will meet on a regular basis in 2025. The members for 2025 include:

- Jenn McNary
- Glenda Thomas
- Tai Pasquini
- Yue Huang
- Representative Jay Livingstone

The working group's statement of purpose is to identify and track legislation and other federal, state (including state agencies), and municipal policies that may impact the rare disease community from a medical, social, economic, or healthcare/service access perspective and to increase awareness of such legislation and policies through education and outreach.

Economic Burden of Rare Disease

The council decided to begin addressing two important legislative charges of the council:

- *Develop methods to publicize the profile of the social and economic burden of rare diseases in the Commonwealth to ensure that the public and health care providers are sufficiently informed of the most effective strategies for recognizing and treating rare diseases.*

- *Determine the human impact and economic implications of early treatment of rare diseases versus delayed or inappropriate treatment of rare diseases as it pertains to the quality of care, the quality of patient's and family's lives, and the economic burdens, including insurance reimbursements, rehabilitation, hospitalization, and related services, on patients, families, and the Commonwealth.*

In the fall of 2024, the council began work on these important focus areas. Council member Tai Pasquini presented a summary of work conducted by the EveryLife Foundation for Rare

Diseases (https://everylifefoundation.org/). The foundation released a study in partnership with the Lewin Group in 2021 to report on the national economic burden of rare diseases in the United States. This report revealed that the economic burden of 379 rare diseases in the United States reached approximately $966 billion in 2019. This estimate surpasses the economic burden of many other chronic diseases, such as diabetes, heart disease, and cancer, according to the CDC. The study, titled "The National Economic Burden of Rare Disease Study," is the most comprehensive assessment, combining data from direct medical costs and previously unexamined indirect and non-medical expenses.

The study also revealed that in 2019, direct medical costs, including hospital visits and prescription medications, amounted to $418 billion, while indirect and non-medical costs, such as lost productivity and non-insured healthcare services, totaled $548 billion. The study highlights the significant financial, medical, and emotional challenges faced by individuals and families affected by rare diseases.

The study also emphasized the need for increased investment in research, awareness, and access to rare disease treatment, labeling them a significant public health crisis. Conducted with the help of multiple stakeholders, including government, industry, and academia, the study's findings were based on a combination of claims data from Medicare, Medicaid, and private insurance, as well as a large survey of the rare disease community. While the study provides valuable insights, its authors caution that the findings represent a conservative estimate and do not encompass all rare diseases, highlighting the need for further research and policy action to address the gaps in care and support for those affected by rare diseases.

The council plans to create a subcommittee in 2025 to investigate the most efficient and effective way to estimate Massachusetts's economic burden of rare disease.

Community Engagement

The council continues to seek new ways to engage with the rare disease community. The goal of community engagement is to learn more about the challenges and needs of those affected by rare diseases in Massachusetts. Council members have discussed ways to connect with the rare disease community for the past two years. In 2024, the council developed and published an online survey (Appendix B) to learn more about the needs of the rare disease community. This survey was available through the Massachusetts DPH RDAC website. The survey had few respondents. The RDAC developed plans to improve the RDAC website to increase visibility in the rare disease community. Additionally, the RDAC had conversations about hosting a series of regional town halls to increase community engagement. In 2025, the council plans to create an official standing subcommittee to address community engagement challenges.

Funds

Accounting of all funds received by the council and the source of those funds:

The Department is working with other appropriate state agencies to establish an account for the RDAC to accept funds.

Accounting of all funds expended by the council:

No funds have been expended.

Summary of RDAC Meetings
The RDAC full council and the steering committee have an alternating, bimonthly meeting schedule. All meetings are open to the public. The council also held workgroup meeting to further understand the topics of community engagement, legislation and how it may impact the rare community and newborn screening. Additional information detailing the discussions held, including the minutes of the meetings as well as any meeting materials presented at the meeting, can be found (https://www.mass.gov/orgs/rare-disease-advisory-council) on the RDAC webpage.

Below is a summary of the meetings in 2024.

Full Council Meetings
The full council meets every other month on the third Thursday from 9:00 am to 11:00 am via ZOOM. A short summary of the meetings is detailed below:

January 18, 2024
The meeting started with a speaker on rare disease. Rachel Bennet, an advocate and parent of a child with a rare disease, spoke to the council about her journey as a parent of a child with visual impairment. She spoke about cerebral visual impairment (CVI) as a leading cause of blindness in children. She voiced her thoughts about adding vision tests to early childhood screening, like hearing screening, noting that early diagnosis of CVI can make a substantial difference when diagnosed early. The rest of the meeting focused on setting priorities for 2024. The council voted to focus on expanding the work of gathering information from the rare disease community, increasing awareness about the RDAC, and tracking and monitoring legislation relevant to the rare disease community.

March 28, 2024
The council welcomed a new member, Representative Joe McKenna, the co-author of the legislation that created this council, replacing Representative Hannah Kane. The guest speakers for this meeting were Sheilah Gauch and Jennifer Vitelli from the PANDA/PANS advisory committee. Sheilah spoke as a parent of two children with a rare disorder. She spoke about the importance of accurate diagnosis. She talked about the confusion between mental health diagnosis and rare disease diagnosis. Once her children were properly diagnosed and treated, they became productive college students.

The next guest speaker, Carolyn Sheridan from the National Organization for Rare Disorders (NORD), spoke about the organization's state report card for Massachusetts. She highlighted four areas in which Massachusetts scored less than an A: medical nutrition, Medicare financial eligibility, newborn screening, and telehealth.

May 16, 2024
The council opened the meeting with a speaker, Dr Michelle Rengarajan. She is the parent of two children diagnosed with the rare disease Duchene's Muscular Dystrophy (DMD) and is an advocate for newborn screening. She spoke about the challenges her first son faced because he was not diagnosed until age 4. Her second son, by contrast, was diagnosed early because his older brother was diagnosed. Most boys with DMD are not diagnosed until 8 to 10 years of age. She saw firsthand why it is important to diagnose this disease early. Her younger son had access to important treatments and therapies that dramatically improved his ability to be independent.

She stated that newborn screening could diagnose this disease at birth, allowing parents time to digest the diagnosis and plan for therapies and treatments that will improve their quality of life. She advocated for adding DMD to the newborn screening test panel.

The council then welcomed NORD representative Allison Herrity to explain the NORD State report card for newborn screening. Allison stated Massachusetts received a B on this record category.

Allison explained the scoring criteria.

- Screening for RUSP core conditions – NORD believes that all states should screen for all the core conditions on the RUSP (The Recommended Uniform Screening Panel is a list of disorders that the US Secretary of the Department of Health and Human Services recommends for states to screen as part of their state universal newborn screening (NBS) programs. Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP.)
- Adding RUSP Core Conditions—NORD believes that states should have a plan to add new RUSP conditions.
- NBS Program Funding – NORD supports policies that state NBS programs should control their own funding
- NBS Advisory Committee – NORD strongly supports the establishment of an advisory committee comprised of diverse stakeholders with a vested interest in NBS.

The next speaker was Dr. Anne Comeau, Deputy Director of the NE Newborn Screening Program, who explained how the program works in Massachusetts. She explained that Massachusetts is different from most states. She explained that the Massachusetts program screens for a core set of disorders and has another set of disorders on a pilot program (which is optional for parents). The advisory committee makes recommendations to the Commissioner, who has the final say on whether a condition is added to the core universal screening panel. The committee consists of a diverse group of stakeholders and evaluates data to determine whether a disorder should be recommended to be added to the core panel.

Several questions were raised, and there was limited time to discuss them. The council decided to have a follow-up meeting to discuss this topic further.

July 18, 2024

The council opened the meeting with a welcome to a new member of the council, Dr. Heather Gray-Edwards. Dr. Gray-Edwards is replacing Dr. Michael Green, who passed away in 2023. The council then heard from Lesa Brackbill, a mom and advocate for newborn screening. Lesa talked about her rare disease journey. Her child, Victoria, was born on July 30, 2014, as a seemingly healthy little girl. However, on February 13, 2015, she was diagnosed with a rare disease called Krabbe Disease. She talked about the words that doctors told her next. If this were caught during newborn screening, she could have been treated. Victoria passed away on March 27 of, 2016. After Victoria's death, Lesa realized that she needed to advocate for newborn screening so that no other parent would go through what she did. She stated that if Victoria had

been born three hours north of her in New York, her life would have looked much different. She began her advocacy work in Pennsylvania (PA), and Krabbe was added to the PA newborn screening panel in 2021. Lesa expressed her passion that babies should not be screened differently just because of the state where they were born. She expressed her willingness to work with the Massachusetts NBS program to make improvements for meaningful change.

The council then heard from Dr. Olaf Bodamer, a physician-scientist, board-certified geneticist, and educator at Boston Children's Hospital, who will soon be a council member. Dr. Bodamer spoke about his work across the globe, which is related to newborn screening. He emphasized the importance of screening, but he cautioned that this was a risk assessment and not a diagnostic tool. He stated that approximately 2/3 of the families seen in his clinic for a positive NBS, need to be made aware of the scope of the NBS Program or their pilot program. He stated that most parents know that their infant received a blood test, but do not know what for. He advocated for better communication with families about to give birth, birthing centers, and healthcare providers about the NBS Program. He touched on the healthcare workforce shortages and the need for increased access to diagnostic testing and treatments. Dr. Bodamer also encouraged the council to consider health equity in their considerations for recommendations.

A panel from the Massachusetts Newborn Screening Program followed Dr. Bodamer. The chair of the Newborn Screening Advisory Committee and other program members were on the panel. Dr. Karen McAlmon, Chair NBSAC Dr. Anne Comeau, Assistant Director NENSP, H Dawn Fukuda, Assistant Commissioner, Bureau Director BIDLS, and Jim Ballin, Office of the General Counsel were members of the panel.

The panel explained the role of the NBS Program as a public health program and the difference between its mandated panel and pilot studies. Dr. McAlmon explained its process and some of its challenges. She explained that the committee lost some members during COVID and was in the process of rebuilding. The panel then answered questions from the council, finishing with a desire to work with the RDAC as it rebuilds and revisits its operating procedures.

September 19, 2024
The council meeting opened with rare disease speaker, Katherin Joyette. Katherin lives in Raynham, Massachusetts. She was diagnosed with a rare condition called small bowel volvulus, which led to a small bowel transplant in 2007. Following years of recovery, the transplant was removed, and she has been reliant on medical nutrition treatment ever since. She expressed that she has not had issues getting her medical nutrition paid for because she has very good private insurance; however, she knows this has not been the case for others.

The next speaker was Hayley Mason from NORD, the National Organization for Rare Disorders. Hayley is a policy analyst responsible for the NORD state report cards for medical nutrition. She discussed the definition of medical nutrition and described why Massachusetts received a B grade. The primary reason for the B grade was limitations on insurance coverage. She explained that two states in the country had an A grade.

Next, the council heard from a dietician, Alison Creeden Griffin who works for New England Life Care, soon be a council member, to speak about her experiences with medical nutrition for

those with a rare disease. She explained that some barriers to accessing medical nutrition included insurance coverage and lack of provider and dietician coverage to oversee the treatment. After some questions from the council members, it was decided that the council needed further discussion. Further discussion and investigation are required to determine if there are significant issues and any issues that the RDAC could make recommendations for improvement.

The council meeting ended with a legislative summary from Representative Livingstone. He presented three bills that recently passed into law that may be of interest to the rare disease community. He presented an overview of the Maternal Health Bill, the Long-term Care Facilities Bill, and the Housing Bond Bill. The meeting ended without definitive next steps for follow-up on medical nutrition. Dr. Tierney stated that he would discuss the next steps with the steering committee.

November 21, 2024
The council meeting opened with rare disease speaker Jessica Souza. Jessica talked about her rare disease journey and focused on the unexpected financial burden caused by her diagnosis. She detailed very specific out-of-pocket expenses and the hardship they have caused. She added that these costs were exacerbated by her forced early retirement.

The next speaker was the DPH Commissioner, Dr. Robbie Goldstein. Commissioner Goldstein spoke to the council about his shared interest in improving the lives of those impacted by rare disease in Massachusetts and offered to work in partnership with the council to assist in its mission. He expressed his appreciation to the members and thanked them for their service to the Commonwealth.

The Commissioner was followed by council member Tai Pasquini, who spoke about the economic burden of rare disease. She summarized a study by the EveryLife Foundation, a national rare disease organization. The study was published in 2021 and detailed the economic burden of rare disease in the United States. Tai facilitated a discussion with the council about ways to estimate the economic costs of rare disease specific to Massachusetts. All agreed that it was an important topic and agreed to make it one of the focus areas for the council in 2025. Tai suggested creating a workgroup subcommittee to investigate the most efficient and effective way to achieve the desired result.

<u>Steering Committee Meetings</u>
The steering committee meets bimonthly (alternating months relative to the full council meetings) to provide leadership and guidance for the council's work. It meets on Thursday mornings from 10:30 to 11:30 a.m. via Zoom.

The RDAC Steering Committee advises the chair on the following topics:

- Agenda setting recommendations
- Identifying recommended meeting speakers
- Recommendations on Council Priorities

The steering committee consists of seven members and is chaired by the RDAC chair. Members include the RDAC chair, a person living with a rare disease, a caregiver or parent of a person living with a rare disease, a legislator, a hospital administrator, a clinician, and a researcher.

Steering Committee Membership (December 2024)

1. RDAC Chair – Dylan Tierney, MD
2. A person living with a rare disease – vacant
3. A caregiver or parent of a person living with a rare disease- Jenn McNary
4. A legislator – Representative Jay Livingstone
5. A hospital administrator – Ryan Thompson, MD, MPH
6. A clinician – vacant
7. A researcher – Tai Pasquini, MPA, PhD

February 19, 2024
This meeting was canceled.

April 11, 2024
The chair opened the meeting by thanking Representative Hannah Kane for her leadership on the council, then welcomed Representative Jay Livingstone as her replacement on the steering committee. He also thanked Dr. David Miller and Guadalupe Hays-Mota for their time and participation on the council's steering committee. He explained that the steering committee would be in flux as members transitioned off and new members joined. Jenn McNary then gave an update on the community engagement workgroup. She expressed the need for more participation in the workgroup and encouraged new members to join. She stated that the workgroup had solicited volunteer help from marketing colleagues to help increase awareness of the RDAC's work and elicit information from the rare disease community to better understand their needs. She hoped to have some recommendations from these outside marketers soon. She then updated the committee on the work of the Legislative Initiatives workgroup. She added that this was a new workgroup whose goal was to identify legislation that may impact the rare disease community. She explained that the workgroup hoped to track legislation and identify meaningful legislation from other states. She asked if the workgroup could weigh in on specific legislation to inform or educate the rare disease community about its existence.

Other members expressed a desire to track legislation, as it would be important to know when making recommendations in our annual report. The committee then went on to identify its priority areas for 2024. Based on the NORD report card, the committee agreed to make newborn screening, medical nutrition, telehealth, and access to specialty providers the priorities for the council in 2024. The committee then discussed the timeline and approval process for its annual report.

June 20, 2024
This steering committee meeting opened with a debrief of the last full council meeting. The primary topic of that meeting was newborn screening, and the committee agreed that more information was needed. Speakers at the last full council meeting focused on the laboratory and technical part of newborn screening. Although this was important to understand, the steering

committee felt the entire council needed to hear from the Newborn Screening Advisory Committee (NBSAC). The NBSAC oversees the NBS as a program. The steering committee discussed inviting the chair of the NBSAC to the next full council meeting. Dr. Tierney said he would invite Dr. McAlmon, the chair of the NBSAC, to attend the July full council meeting. He then asked the council about the topic for the September meeting. All agreed that Medical Nutrition would be the topic, and possibly telehealth if we have more information.

August 15, 2024
This meeting opened with a discussion on the changes in council membership and, therefore, the changes coming for the steering committee. Dr. Tierney welcomed Tai Pasquini, the newest member of the steering committee. Dr. Tierney then summarized the discussion of the ad hoc newborn screening meeting held last month. This ad hoc meeting aimed to develop recommendations on the Newborn Screening program to be included in the RDAC annual report. Dr. Tierney shared the following recommendations from this meeting.

General Recommendations

1. Increase transparency
2. Update the website to make as much information as possible available to the public
3. Ensure compliance with Open Meeting Law
4. Reevaluate the education materials for NBS. Consider including webinars, videos, and other modes of communication to explain the program. Simplify the parental brochure according to health literacy, ADA, and plain language principles.
5. Consider broader definitions of treatment benefits to be used as criteria for acceptance or rejection of a given condition for adoption on the Massachusetts newborn screening panel
6. Investigate any new technologies to modernize the program annually and report findings to the DPH commissioner

The steering committee discussed these recommendations and added the following.

- Add a regulation change for the title by removing the word "blood" from the regulations' title allowing for the modernization of the screening as procedures as appropriate.
- Add the word “diagnosis” to the benefit. The discussion focused on realizing the diagnosis benefit as well as the treatment benefit part of the recommendation.
- Add more clarity around transparency. What transparency would we like to see?
- Provide more clarity around the process for reviewing new conditions. What would that look like if a condition were added to the RUSP? What would that process look like if a new condition, not on the RUSP, was nominated?

Dr. Tierney agreed to review and revise the recommendations before the next full council meeting. Jenn McNary then gave an update from the Legislative Initiatives workgroup. She let everyone know that the group had developed goals and objectives. She stated that she had been reaching out to other RDACs in other states, and some took a more active role in legislative actions. She hoped to get clarity from the DPH legal counsel before the next meeting in mid-September. Dr. Tierney said he would send their document to the legal team for review. The rest of the meeting included the RDAC annual report, timeline, and review process.

October 17, 2024
Dr Tierney opened the meeting by discussing RDAC operations and DPH oversight of the council. Dr. Tierney stated that he has been meeting with DPH leadership and the DPH general counsel and had some updates. He started by saying that the legal counsel reviewed the council's legislative language, and they believe that the words "*There shall be a rare disease advisory council within the department*" means that the RDAC is a council of DPH and under the direction and control of DPH. He added that the Commissioner's office would be more active in setting agendas and priorities with mutual DPH and RDAC priorities in mind. He plans to regularly meet with the BIDLS leadership, specifically Dawn Fukuda, about the council. Dawn Fukuda will meet with the Commissioner regularly and will be the liaison between the Commissioner and the council chair.

He then added that the general counsel let him know that our subcommittees and working groups must follow open meeting law. They also stated that any subcommittee/work group of the council must have a defined odd number of members and a defined goal and must be voted on by the full council. They had some specific questions about the legislative subcommittee and suggested that Thera Meehan, BIDLS Director of Policy, track any bills relevant to the RDAC. He added that DPH cannot comment on any legislation. As a council under the direct guidance of the Commissioner, the council could not comment on the pros or cons of any specific legislation. He stated that DPH wants to have control over any information specific to legislation. He noted that the RDAC needed to develop a specific structure for how they will be involved in any specific legislation, and he will bring it back to DPH for approval on moving forward or not.

After much discussion with steering committee members, the most significant question was whether DPH had veto powers over the council's agendas and recommendations. Dr. Tierney could not answer the question, but he stated that he would follow up to find out.

The rest of the meeting included a review of the newborn screening recommendations. Dr. Tierney presented a streamlined version of the recommendations the newborn screening ad hoc workgroup presented. Members had many questions about what was presented. Dr. Tierney stated that the committee would like to meet again before the next full council meeting due to a need for more time for more discussion. All agreed. Representative Livingstone asked if the Commissioner could meet with the steering committee. Dr. Tierney stated that he would bring the request back to DPH. All agreed to meet again on October 31 from 11:30 to 12:30 to continue the discussion.

October 31, 2024
Dr. Tierney opened the meeting by introducing Dawn Fukuda. Dawn is an assistant commissioner and is responsible for overseeing this council. She commended the council for its work to date and stated that the Commissioner's office fully supported their work. She stated that DPH would be a partner with the council. The relationship will be supportive. She expressed that the Commissioner would like to know the council's priorities ahead of time so that he could provide DPH context if possible. She further expressed that DPH did not control the council's agendas. However, the Commissioner's input could strengthen the work of the council. She answered members' questions and clarified that DPH does not have veto powers over the council. She did state that it would be helpful if the council and DPH had aligning positions.

Dr. Tierney then introduced steering committee member Tai Pasquini, who talked about one of the council's legislative charges related to the economic burden of rare disease. She discussed the study conducted by Every Life Foundation and her discussion with the company that conducted that study. She presented a proposal from them to conduct a similar study specific for Massachusetts. After many questions, it was agreed to present this to the full council for further discussion. Tai agreed to present this topic at the November full council meeting.

Workgroups

COMMUNITY ENGAGEMENT WORKING GROUP

April 26, 2024
The Community Engagement Workgroup (CE) held its first meeting to discuss the goals and objectives of the committee. The committee is co-chaired by council members Jenn McNary and Glenda Thomas. Jenn welcomed some marketing experts from the industry to speak about ways that the RDAC could increase awareness and improve its interactions with the rare community and its stakeholders. The group discussed ways to accomplish this without a budget. The committee ended with offers from the marketing guest speakers to develop a marketing plan for the council and share it with the chairs. They talked about social media presence and improving the RDAC website.

May 15, 2024
This meeting focused on the marketing plan created by industry marketing experts. The group discussed how some goals and objectives were pertinent and others were not. They welcomed the DPH IT person to explain how the RDAC website could be updated while remaining in compliance with ADA standards. The committee agreed to work on edits and content to redesign the website to make it more user-friendly. They felt there needed to be more pictures and more engagement from the users. They also discussed adding videos and how that may be possible.

The council will discuss forming a standing subcommittee to address community engagement at the next full council meeting.

LEGISLATION AND POLICY WORKING GROUP

April 22, 2024
The Policy Workgroup held a meeting to discuss its goals and objectives. Jenn McNary and Glenda Thomas co-chair the committee. The group discussed and agreed on its goal of identifying how existing and future legislative initiatives may address issues facing the rare disease community. They next discussed their objectives to create a tracking form to follow the key areas of medical nutrition, newborn screening, telehealth, and access to specialty care, including home care. They discussed creating a worksheet for tracking, deciding on keywords for the search, and identifying resources to help with this work. They discussed talking with other state RDACs to see what they are doing about legislative initiatives. The group agreed that they would not be able to advocate for any of the legislation that was identified and wanted to focus their energy on informing and educating about the legislation. They ended by asking for

some legal consultation to better understand what they could and could not do related to legislation.

May 6, 2024
This meeting focused on finalizing the workgroup's goals and objectives. All agreed to accept the goals and objectives as presented. One member shared that five other RDACs in other states have a legislative committee, and she would reach out to them to see what they are working on. Representative Livingstone walked through the legislative process for the committee. He stated that it was important to understand the timeline of how legislation moved through the process. He reiterated that the RDAC could support bills important to the rare community by providing education and testimonials. Representative McKenna stated that even though the RDAC could not advocate for specific legislation, it could be very helpful in providing education and resources. He recommended that the RDAC hold learning sessions for the legislature to better understand the issues and the impacts on the rare community. He stated that many nonprofits come to the statehouse to provide education and testimonials about a specific topic. He also emphasized that bills may take many iterations before becoming law. They often change names and numbers and are sometimes difficult to follow. Representative Livingstone offered his staff and interns to help with this process. He also added that the statehouse news often has valuable information about specific bills.

May 20, 2024
This meeting of the committee focused on identifying and finalizing the keyword search for legislation. Several members contributed thoughts based on the identified focus areas of the council. The keywords agreed to included newborn screening, diagnostic testing, access, diagnosis, coverage, MassHealth Connector, health savings account, home care, person care attendant program, person care management, complex care management, rare disease, physical therapy, special education, telehealth, MassHealth, medical nutrition, durable medical equipment, housing for the disabled, supplemental income, rehabilitation, specialty care, out of network care, orphan drugs, gene therapy, drug utilization, pharmacy and therapeutics, medical transportation, mental health and caregivers.

Next, the committee discussed the criteria by which it would prioritize its research. J McNary presented the following criteria: (1) it must impact the rare community, and (2) it is currently an active bill.
Representative Livingstone then updated the committee on the status of the current healthcare bill.

June 3, 2024
This meeting focused on finalizing the keywords used to search for legislation. They also heard from a DPH guest speaker, Thera Meehan, who tracks legislation as part of her scope of work. She expressed that she could create a spreadsheet for the committee based on their search words but that legislation is often hard to follow. She explained that bill numbers change, sometimes bills merge with other bills, and sometimes they change completely through the process. Members asked about getting bill summaries. Representative Livingstone offered to share any

bill summary that came to his office. He also shared that this was the end of the legislative session so any bills they are currently tracking may never move forward. He suggested starting the committee process in January. He also reiterated that the committee should focus on educating, increasing awareness, and not advocating. He suggested getting legal advice about what the committee could do.

July 15, 2024
This meeting focused on finalizing the legislative tracking spreadsheet, including how it would be set up and monitored. Representative Livingstone's office shared a sample of the spreadsheet they use to track bills and other legislation. It became clear that tracking all the keywords would be overwhelming. The committee discussed how it would choose what to follow. Representative Livingstone offered to have his staff work on a spreadsheet sample for the committee. All agreed that it would be way too much work to review everything. The committee members do not have the capacity to do this type of research. All agreed that the committee needed some legal guidance before moving forward. Dr. Tierney offered to reach out to DPH legal for some guidance.

The council formalized this workgroup as a standing subcommittee at the November full council meeting. The subcommittee members include:

Chairs: Jenn McNary and Glenda Thomas
Representative Jay Livingstone
Yue Huang
Tai Pasquini

The subcommittee has decided to meet quarterly in 2025.

Plans for 2025

In 2025, the RDAC hopes to continue building on its progress to better understand the needs of the rare disease community. The council will continue its work on medical nutrition by trying to better understand the specific challenges related to medical nutrition access for those with a rare disease. The council will focus on community engagement and identifying the ways to reach the rare disease community in Massachusetts. The newly formed policy subcommittee will continue to learn about legislative actions and how they may impact the rare community and how to keep the public informed. Addressing the economic burden of rare disease in Massachusetts will take enormous effort but the council is committed to begin this work. The discussions on how to tackle this legislative mandate will begin early in the year. The council plans to expand awareness about its work through an enhanced online presence and more active social media engagement. With a better understanding of the needs of people with rare diseases, the RDAC will be able to develop additional recommendations for the Massachusetts legislature about how to improve the lives of people with rare diseases in the Commonwealth in the coming year.

Conclusion

Massachusetts Rare Disease Advisory Council is creating a more supportive environment for individuals with rare diseases and improving their access to necessary medical care, treatments, and social supports. Although the council has more work to do, it is focused on increasing awareness and improving collaborations, leading to more effective strategies for managing rare diseases in the Commonwealth.

Appendix A: Rare Disease Advisory Council Membership – December 2024

	Seat	Member	Appointed By
1	Commissioner, DPH, or designee	Dylan Tierney, MD, MPH	Commissioner, DPH
2	Executive Director, Health Policy Commission, or designee	Yue Huang, MS	Executive Director, Health Policy Commission
3	Senate Member 1	Senator Paul Feeney	Senate President
4	Senate Member 2 (Minority Leader)	Tai Pasquini, MPA, PhD (designee of Senator Bruce Tarr)	Senate Minority Leader
5	House Member 1	Representative Jay Livingstone	Speaker of the House
6	House Member 2 (Minority Leader)	Representative Joseph McKenna	House Minority Leader
7	Pharmacist – Experience with Rare Disease Drugs	Shivang Patel, PharmD	Senate President
8	Geneticist	Diane Lucente, MS., LCGC	Senate President
9	Nurse with Experience Treating Rare Disease	Lena Joseph, RN, CPN	Senate President
10	Senate President 4	VACANT	Senate President
11	Representative of a Health Plan	Gail Ryan, PharmD	Speaker of the House
12	Genetic Counselor with Rare Disease Experience	Asma Rashid, MS, CGC	Speaker of the House
13	Representative of a Rehab Facility	Ross Zafonte, DO	Speaker of the House
14	House Speaker 4	VACANT	Speaker of the House
15	Academic Research Institution 1	Heather Gray-Edwards, DVM, PhD	Governor
16	Academic Research Institution2	Andrew Dwyer, PhD, FNP-BC	Governor
17	Physician with Rare Disease Experience 1	Olaf Bodamer, MD, PhD FACMG	Governor
18	Physician with Rare Disease Experience 2	Christelle Achkar, MD	Governor
19	Hospital Administrator 1	Ryan Thompson, MD, MPH	Governor
20	Hospital Administrator 2 - Pediatric	Charlotte Boney, MD	Governor
21	Representative of Rare Disease Organization 1	Julie Gortze, RN	Governor
22	Representative of Rare Disease Organization 2	VACANT	Governor
23	Representative of Rare Disease Organization 3	Jenn McNary	Governor
24	Biotechnology and Scientific Community 1	Jeff Livingstone	Governor
25	Biotechnology and Scientific Community 2	Robert E. Schultz, MBA	Governor
26	Dietician -experience with Rare Disease	Ann Wessel,	Governor
27	18 or older with Rare Disease 1	Glenda Thomas	Governor
28	18 or older with Rare Disease 2	VACANT	Governor
29	Rare Disease Caregiver	Alexsandra Mahady	Governor

Appendix B: Rare Disease Advisory Council Community Engagement Survey

Survey Link: https://docs.google.com/forms/d/e/1FAIpQLScCwos-UKoRMe7_UOQ9x4F0ThXTFvDD-USPlhRQMaWV5eneCQ/viewform

Form Preface - Survey Greeting Page

About RDAC
The Rare Disease Advisory Council was established by an act promoting a resilient healthcare system that puts patients first (Section 26 of Chapter 260 of the Acts of 2020).

RDAC mission statement
The Massachusetts Rare Disease Advisory Council's mission is to provide guidance and recommendations on rare disease incidence and the status and needs of the rare disease community to the governor, the legislature, and the Department of Public Health, with the goal of improving the lives of people impacted by rare diseases in the Commonwealth.

Strategic Priorities:

1. Improve healthcare access and quality of care for people with rare diseases.
2. Advocate for and improve access to social supports and services for people impacted by rare diseases.
3. Foster communication and collaboration to empower the rare disease community in Massachusetts.

Objective of This Survey: The Massachusetts Rare Disease Advisory Council's mission is to provide guidance and recommendations on rare disease incidence and the status and needs of the rare disease community to the governor, the legislature, and the Department of Public Health to improve the lives of people impacted by rare disease in the Commonwealth. This survey should take less than 10 minutes, but the information given can change the future of rare disease patients in the Commonwealth of Massachusetts. The council is gathering information on the resources currently available to rare disease patients and caregivers and resources that are difficult to obtain or not available. We are also interested in learning who the stakeholders are and what advocacy groups we can reach out to when we have questions. Your input is critical to the success of this survey. Results will be aggregated and summarized in a council report. WE NEED YOUR HELP. Please forward this to anyone you think would help complete the information.

Please note: All information provided is confidential, and your participation is voluntary. However, we can't emphasize enough how important it is that we hear from you!

1. *Please select which portion of this form you'd like to access:
    - ○ Section 1 - Support Resources

○ Section 2 - Contact Information and Affiliation

Note: Upon answering the Form Pre-Face question above the user will either be directed to Section 1 or Section 2. If the user chooses to, they may only elect to complete Section 2.

Section 1 - Support Resources

Please note that many questions regarding your identity are entirely optional to enter.

1. Your first name:_____
2. Your last name:_____
3. Your contact E-Mail:_____
4. Are you entering this as an individual or as a representative of an organization?
    - ○ I am entering this information as an individual
    - ○ I am entering this information as a representative of an organization
5. Phone number:_____
6. *Name of Massachusetts rare disease social support resource:_____
7. Website for Resource:_____
8. Contact Information for Resource:_____
9. *Type of Resource:
    - □ Medical
    - □ Personal care service
    - □ Housing
    - □ Social, emotional, and recreational
    - □ Financial
    - □ Insurance and healthcare coverage
    - □ Other:_____

10. *Age of Population Served:

□ 0 years to 12 years

□ 13 years 17 years

□ 18 years to 21 years

□ 22 years 65 years

□ 65 and older

□ All ages

11. *Disability Status of Population Served:

○ Disabled

○ Not disabled

○ Disability status not relevant

○ Other:_____

12. *Disease Diagnosis of Population Served (e.g., autoimmune pulmonary alveolar proteinosis, general rare disease, undiagnosed, etc.):_____

13. *In which Massachusetts county/counties are the rare disease support services based?

□ Barnstable County

□ Berkshire County

□ Bristol County

□ Dukes County

□ Essex County

□ Franklin County

□ Hampden County

□ Hampshire County

□ Middlesex County

□ Nantucket County

□ Norfolk County

□ Plymouth County

□ Suffolk County

□ Worcester County

14. *Have you accessed this resource?

○ Yes

○ No

15. If you accessed this resource, in what year?

□ Within the past year

□ Within the past 1-3 years

□ Within the past 3-5 years

□ More than 5 years ago

16. *If you accessed this resource, where did you live?

□ Barnstable County

□ Berkshire County

□ Bristol County

□ Dukes County

□ Essex County

□ Franklin County

□ Hampden County

□ Hampshire County

□ Middlesex County

□ Nantucket County

□ Norfolk County

□ Plymouth County

□ Suffolk County

□ Worcester County

17. What resources do you wish you had access to that you do not currently have?:_____

18. *Would you like to list another support resource? (Please Note: you may submit up to 10 resources)

○ Yes

○ No

Note: Upon answering yes, the user will be directed to an alternative version of Section 1 that will ask exactly the same questions as #4-16. This process will continue until the user selects "No" or 10 resources have been submitted. Following this the user will be prompted to complete Section 2

Section 2 - Contact Information and Affiliation

1. Which of the following groups do you personally represent? Check all that apply

□ Legislator

□ Pharmacists/specialty pharmacy

□ Dietician

□ Geneticist

□ Nurse/Nursing organization

□ Health insurance plan

□ Genetic counselor

□ Rehab facilities

□ Academia research institution

□ Physician

□ Hospitals

□ Patient groups

□ Biotech and scientific community

□ Caregivers

□ Entrepreneurs associated with rare disease

2. First Name:_____

3. Last Name:_____

4. Phone Number:_____

5. Fax:_____

6. Email:_____

7. Business Address:_____

8. Healthcare Profession:_____

9. Any affiliation to any other organization:_____

10. Is this organization private or nonprofit?

○ Private
○ Nonprofit
○ Other:_____

11. Organization website:_____

12. Disease of interest:_____

13. Medical institution:_____

14. Your organization's goal / mission:_____

15. Will you be interested in lending your expertise in the future?

○ Yes

○ No

16. Would you like to be contacted for a future event or collaboration?

○ Yes

○ No

Please note any additional comments or feedback that you'd like to add:

_____

Note: Upon answering the final question in Section 2 the user will be able to click the "Submit" button at the bottom of the form. Following their form submission, the user will land on a confirmation page stating:

> "Your response has been recorded. We thank you for taking your time and helping Massachusetts's rare disease advisory council."