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**Annual Report on Establishing a Rare Disease Advisory Council**

**December 2023**

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# Letter from the Chair

December 31, 2023

On behalf of the members of the Massachusetts Rare Disease Advisory Council (RDAC), it is my pleasure to submit the second annual legislative report in compliance with the Massachusetts Acts of 2020, Chapter 260. This report summarizes the collaborative efforts of the RDAC members over the past year.

Over the course of 2023, Massachusetts RDAC focused on gathering and sharing information critical to our mission to make recommendations to the governor and legislature about how to improve the lives of people with rare disease in the Commonwealth. Our bimonthly full council meetings alternating with smaller working group meetings are where this gathering and sharing takes place. As chair of these meetings, I am proud to report that they serve their purpose as a space for learning, for debate and for progress. Our deliberations have clarified the task in front of us: to identify the common experiences (challenges) faced by the estimated population of 200,000 to 700,000 Massachusetts residents with a rare disease. To do this, we have called on people from across the rare disease community. We have heard from legislators and researchers, primary care physicians and health policy experts, social workers and community advocates. Most importantly, we have heard from people with rare diseases.

While we have made progress in 2023, our work in understanding the challenges of rare disease is not done. In the coming year, we hope to extend our reach into the community to capture more fully the range of experiences and opinions of the diverse population of people living with rare disease in Massachusetts. We propose to develop a marketing plan to understand the populations we hope to serve and how best to engage with them in our work. We will also focus on exploring possible solutions to the challenges identified through our conversations, keeping in mind the need to produce true positive impact on the lived experience of persons living with rare diseases, while at the same time developing responses that are feasible to fund and implement.

Once these solutions become clear, we will make our recommendations to the legislature with the confidence that we represent the voice of the community that we have been assembled to serve.

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

# Executive Summary

The Massachusetts Rare Disease Advisory Council (RDAC) is pleased to present its 2023 annual legislative report in accordance with the Acts of 2020 Chapter 260. The RDAC was established as part of Senate Bill 2984, An Act promoting a resilient healthcare system that puts patients first. The legislature enacted the bill in December 2020, signed into law by Governor Charlie Baker on January 1, 2021 (<https://malegislature.gov/Laws/SessionLaws/Acts/2020/Chapter260>).

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 a rare disease in the Commonwealth. This report describes the accomplishments of the RDAC over the past year.

A priority for 2023 was to determine the prevalence of rare disease in Massachusetts. Prevalence estimates will provide the public, the governor, and the legislature with a sense of the magnitude of rare disease in the Commonwealth. Estimating the prevalence, however, is challenging. There are over 7000 rare diseases but few existing registries that count people with a given disorder. Additionally, because rare diseases affect a small number of people, there may be few individuals to count. There are rare diseases that are so poorly understood that they do not even carry a name to be counted.

A workgroup of RDAC members presented a report on prevalence of rare disease in Massachusetts that was approved by the full council in September 2023 (see Appendix A for the full report). The council estimates that between 200,000 to 700,000 people in the Commonwealth of Massachusetts have a rare disease. This state-wide estimate was derived by extrapolating from international and national estimates of rare diseases. The RDAC estimate of prevalence was developed as a range to capture the uncertainty inherent in a calculation that is based on limited primary data. Despite this caveat, the RDAC feels confident that there is a significant number of people in the state who live with a rare disease.

The other major activity of the council in 2023 was to develop a better understanding of the needs of the rare disease community. The RDAC undertook two projects to accomplish this goal. The first was to develop a needs assessment survey to be used to query the community of rare disease stakeholders in Massachusetts. The first draft of this survey was completed and is attached in the appendix.

The RDAC also sought direct comment from people with rare diseases through personal testimonies. A individual living with a rare disease is invited to start every full council RDAC meeting with comments about their perspective as a person living with a rare disease. The RDAC also held a virtual town hall event in October 2023. This town hall allowed community members to provide testimony about their experiences with rare diseases directly to the RDAC. Through these two outlets, people living with a rare disease expressed that the following topics were of the most interest:

* limited access to mental health and social supports,
* limited access to specialty providers (due to scarcity of providers or lack of insurance coverage),
* need for acute to supportive transitional care
* home health care workforce issues,
* importance of telehealth for care delivery,
* burdensome health insurance requirements including copay accumulators, and prior authorization and poor coverage of specialty medications.

Testimony from this event can be found in the appendix.

For the upcoming year, the RDAC hopes to continue to build on it’s progress in better understanding the needs of people with rare disease. We hope to expand the awareness of the community about the RDAC’s work through an enhanced online presence and more active social media engagement. We expect that with better understanding of the needs of people with rare disease, the RDAC will be able to develop specific recommendations for the Massachusetts legislature about how to improve the lives of people with rare disease in the Commonwealth in the coming year.

# Council Membership

The council membership has had a few changes in 2023.

Michael Green, MD, Ph.D., Director of the University of Massachusetts Cancer Center and Co-Director of the Li Weibo Institute of Rare Diseases, who was appointed by the governor and represented an academic research institution that receives grant funding for rare disease research, passed away in February of 2023. We are awaiting an appointment for his replacement.

Michael Sherman, MD, retired from his position at Harvard Pilgrim Health Care in the spring of 2023. He was replaced by Gail Ryan, PharmD, to represent a health plan or ACO certified by the Health Policy Commission. Gail joined the council in August.

Lisa Deck, Founder and President of Sister@Heart the governor appointed as representative from a rare disease patient organization that operates in Massachusetts resigned in November. We are awaiting a replacement.

Celia Segel, designee for the commissioner of the Health Policy Commission, left her position in September and was replaced by Yue Huang, MS.

Below is the current membership of the full council as of December 2023.

|  |  |  |  |
| --- | --- | --- | --- |
|  | 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) | Senator Bruce Tarr (Ms. Tai Pasquini, MPA, PhD) | Senate Minority Leader |
| 5 | House Member 1 | Representative Jay Livingstone | Speaker of the House |
| 6 | House Member 2 (Minority Leader) | Representative Hannah Kane | House Minority Leader |
| 7 | Pharmacist – Experience with Rare Disease Drugs | Shivang Patel, Pharm.D. | 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 | Michele Rhee, MBA, MPH | 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 | Janis Creedon | Speaker of the House |
| 15 | Academic Research Institution 1 | VACANT | Governor |
| 16 | Academic Research Institution2 | Andrew Dwyer, PhD, FNP-BC, FNAP, FAAN | Governor |
| 17 | Physician with Rare Disease Experience 1 | Andrew Lane, MD, PhD | Governor |
| 18 | Physician with Rare Disease Experience 2 | David Miller, MD, PhD | Governor |
| 19 | Hospital Administrator 1 | Ryan Thompson, MD | 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 | Lisa Deck (resigned 11/2023) | Governor |
| 23 | Representative of Rare Disease Organization 3 | Jenn McNary | Governor |
| 24 | Biotechnology and Scientific Community 1 | Jeff R. Livingstone, PhD | Governor |
| 25 | Biotechnology and Scientific Community 2 | Robert E. Schultz, MBA | Governor |
| 26 | Dietician -experience with Rare Disease | Ann Wessel, MS, RD, LDN | Governor |
| 27 | 18 or older with Rare Disease 1 | Glenda Thomas | Governor |
| 28 | 18 or older with Rare Disease 2 | Guadalupe Hayes-Mota, MBA, MS, MPA | Governor |
| 29 | Rare Disease Caregiver | Alexsandra Mahady | Governor |

# Subcommittees

The council established a steering committee in March of 2022. The steering committee meets bimonthly (alternating months relative to the full council meetings) to provide leadership and guidance for the council’s work.

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. The committee is chaired by the RDAC chair, and members include current members of the RDAC and represent the following categories:

1. People living with a rare disease
2. Caregivers or parents of people living with a rare disease
3. The legislature
4. Hospital administrators
5. Clinicians
6. Researchers

The full RDAC voted on Steering Committee membership on March 16, 2022.

Steering Committee Membership

RDAC Chair – Dr. Dylan Tierney

People living with a rare disease – Michele Rhee

Caregiver or parent - Jenn McNary

Legislator – Representative Hannah Kane

Hospital administrator – Dr. Ryan Thompson

Clinician – Dr. David Miller

Researcher – Dr. Jeff Livingstone

Guadalupe Hayes-Mota replaced Dr. Jeff Livingstone on the steering committee in December 2023.

# Workgroups

Three workgroups were added in 2023 to assist the council in information-gathering and data collection.

Workgroup 1, chaired by Dr. David Miller, was established to:

Improve healthcare access and quality of care for people with rare diseases.

GOAL: Determine the prevalence of rare diseases in Massachusetts.

Workgroup 1 Membership

Dr. David Miller (chair)

Alexsandra Mahady

Andrew Dwyer, PhD, FNP-BC, FNAP, FAAN

Andrew Lane, MD, MPH

Tai Pasquini, MPA, PhD

Ann Wessel, MS, RD, LDN

Celia Segel, MPP

Julie Gortze, RN

Ryan Thompson, MD

Jeff Livingstone, PhD

This group met regularly in 2023 and presented their prevalence report to the full council on September 21, 2023. (Appendix A) The prevalence report reflects the work of this workgroup to determine the prevalence of rare diseases in Massachusetts.

Workgroup 2, chaired by Jenn McNary, was established to:

Advocate for and improve access to social supports and services for people impacted by rare diseases.

GOAL: Develop a profile summary of existing and needed rare disease social supports and services in Massachusetts

Workgroup 2 Membership

Jenn McNary (chair)

Lisa Deck

G. Hayes-Mota, MBA, MS, MPA

D. Lucente, MS, LCGC

M. Rhee, MBA, MPH

R. Zafonte, DO

J. Creedon,

Representative Jay Livingstone

This group met regularly in 2023 and continues to gather information to develop a profile of rare disease social supports and services in Massachusetts.

Workgroup 3, chaired by Lena Joseph, RN, CPN was established to:

Foster communication and collaboration to empower the rare disease community in Massachusetts.

GOAL: Develop a profile of rare disease expert individuals, community-based organizations, voluntary organizations, healthcare providers, and any other public or private organizations with an interest in rare diseases in Massachusetts.

Workgroup 3 Membership

Lena Joseph (chair)

Glenda Thomas

Shivang Patel, PharmD

Jenn McNary

Charlotte Boney, MD

Representative Hannah Kane

Robert Schultz, MBA

Asma Rashid, MS, CGC

Senator Paul Feeney

This group met regularly in 2023 and continues to gather information to develop a profile of rare disease expert individuals, community-based organizations, voluntary organizations, healthcare providers, and other public or private organizations with an interest in rare diseases in Massachusetts.

Workgroups 2 and 3 decided to meet together to develop a survey tool for gathering the information for both profiles. The survey was developed (Appendix B) and was launched on October 28, 2023. The workgroups plan to keep the survey open for 3-4 months. Once the data is collected and verified, they will create their respective profiles in 2024.

# Public Town Hall

The council held its first virtual town hall on Saturday, October 28th, to listen and learn from the public and the rare disease community. As the council continues to gather information, it realizes how important it is to hear from individuals about the resources and social supports most important to those living with a rare disease in Massachusetts. The council asked individuals to speak about the resources and social supports that are unavailable or difficult to access in Massachusetts. The information gathered at this meeting will help the council better understand the needs of people impacted by a rare disease and living in Massachusetts in an effort to make meaningful and impactful recommendations to the governor, the legislature, and the Department of Public Health. Summary of the event (Appendix C)

# The Council’s Purpose

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

1. 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 for the purpose of ensuring greater cooperation regarding the research, diagnosis, and treatment of rare diseases. This coordination may require:
	1. Disseminating the outcomes of the advisory council’s research and identifying best practices and policy recommendations
	2. Utilizing common research collection and dissemination procedures
2. Using existing publicly available records and information to undertake a statistical and qualitative examination of the prevalence and causes of rare diseases in order to develop a profile of the social and economic burden of rare diseases in the Commonwealth.
3. 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.
4. 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.
5. 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.
6. 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.
7. 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.
8. Examine the feasibility of developing a rare disease information and patient support network in the Commonwealth to aid in determining any genetic or environmental contributors to rare diseases.
9. 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, and 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 any gifts, donations, grants, bequests, or federal funds, for the purpose of carrying out the above charges.

# 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, and 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 2023

January 19th, 2023; Full Council Meeting

Workgroups were established at this meeting.

Workgroup 1 would focus on determining the prevalence of rare diseases in Massachusetts.

Workgroup 2 would focus on developing a profile of rare disease social supports and resources in Massachusetts.

Workgroup 3 would focus on developing a profile of rare disease expert individuals, community-based organizations, voluntary organizations, healthcare providers, and any other public or private organizations with an interest in rare diseases in Massachusetts.

February 16th, 2023; Steering Committee Meeting

This meeting focused on planning for Rare Disease Day at the Statehouse on February 28th

Discussions began on the planning for a virtual town hall in the fall.

March 16th, 2023; Full Council Meeting

The council welcomes Senator Paul Feeney to the council. The council received word of the passing of a council member, Dr. Michael Green. His obituary was shared with the council. Each workgroup provided an update on their progress. The National Organization of Rare Disorders presented an overview of their organization.

April 27th, 2023; Steering Committee Meeting

The committee received an update from each workgroup chair and then discussed plans for a virtual town hall in the fall.

May 18th, 2023; Full Council Meeting

After an update from each workgroup chair, the council heard from MassHealth Office of Long-Term Services and Supports, which provides a robust system of care for members of all ages who need services to enable them to live with independence and dignity in their daily lives, participate in their communities, and increase their overall quality of life.

June 8th, 2023; Steering Committee Meeting

The committee discussed project ECHO, a virtual learning platform for rural providers to gain expertise on a specific topic or specific disease. The committee discussed a possible partnership with Project ECHO to help rural providers gain expertise on rare diseases. Planning for the public town hall continued.

July 20th, 2023; Full Council Meeting

The council heard from two rural providers who care for patients with rare diseases to gain a better understanding of the challenges they face caring for complex patients in a rural setting. Workgroup 1 presented a draft of their prevalence report.

August 17th, 2023; Steering Committee Meeting

The committee heard from one of the authors of the Autism Report presented to the state legislature in 2021. Discussions focused on plans for developing recommendations and how to present them to the legislature. As an action-oriented council that is only advisory in nature, the committee wants to be strategic in developing its recommendations. Other discussions were related to tracking legislation that may be important to the rare community. What would the council’s role be in this type of activity?

September 21st, 2023; Full Council Meeting

The council welcomed two new members. Gail Ryan, PharmD would replace Dr. Michael Sherman and Yue Huang, MS would replace Celia Segel, as designee from Health Policy Commission. The council heard for the Director of Social Services at Boston Children’s Hospital to learn more about some of the challenges that pediatric patients with a rare disease face, especially with the transitions of care throughout their childhood and into adulthood. Other discussions involved setting the date for the Virtual Town Hall. October 28th was selected as the best date. Workgroup 1 presented its final prevalence paper, and the council approved it. (Appendix A) Workgroup 2 and 3 chairs discussed their final version of a survey tool and plans to release it at the Virtual Town Hall. The survey will be available in English, Spanish, Portuguese, and Chinese. Survey tool (Appendix B)

October 19th, 2023; Steering Committee Meeting

The council attendees of the National Organization of Rare Diseases & Orphan Products Breakthrough Summit summarized the event for the committee. The remainder of the meeting focused on planning and marketing of the Virtual Town Hall to be held on Saturday, October 28th.

November 16th, 2023; Full Council Meeting

The council heard from the Executive Director of the Minnesota Rare Disease Advisory Council. Minnesota has evolved into an executive branch of the state legislature. Their council has decided to focus on four pillars with a priority on rare diseases. Those pillars include increasing the understanding of the collective barriers to care, reducing the time to diagnosis, increasing care coordination, and improving care transitions. This presentation gave the council a lot to think about as they discussed the future of the RDAC in Massachusetts. Workgroups 2 and 3 updated the full council about their survey. They asked the council for help in developing a marketing plan to get the word out about the survey.

December 19th, 2023; Steering Committee Meeting

The steering committee focused on setting priorities for the council in 2024. All agreed that more data and information would be necessary to support the case for an office of rare disease in the state legislature. The committee discussed forming new subcommittees to discuss and gather information on specific topics from the public inquiries in 2023. However, the consensus was to focus on gathering the data needed to send meaningful recommendations to the governor and legislature by the end of the calendar year.

# Rare Disease Advisory Council Accomplishments

The Massachusetts RDAC was established 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 a rare disease in the Commonwealth.

The RDAC has identified activities that will help to establish a better understanding of the burden of rare diseases in Massachusetts:

* to determine the prevalence of rare diseases in Massachusetts,
* to identify the supports and services available to the rare disease community in Massachusetts,
* to identify the stakeholders with interest in the rare disease community in Massachusetts.

As these information-gathering activities are completed, the RDAC will be better positioned to summarize the current state of the rare disease community in the Commonwealth and provide guidance and recommendations as legislated.

Actions taken and progress made toward implementing the comprehensive rare disease plan. Progress toward the development of a comprehensive rare disease plan includes:

* Assembly of the 29-member council
* Establishment of RDAC standard operating procedures
* Formation of the steering committee to guide the RDAC agenda
* Identification of RDAC’s mission, strategic priorities, and goals
* The formation of three workgroups to gather data and information to help the council develop recommendations that are most relevant to the rare disease community
* Consultation with state, federal, and other experts on rare diseases.

An accounting of all funds received by the council and the source of those funds

No account has been set up for the RDAC to accept funds. No funds have been accepted.

An accounting of all funds expended by the council.

No account has been set up for the RDAC to expend funds. No funds have been expended.

To the extent practicable, an estimate of any cost savings on the part of individuals and the Commonwealth will occur upon full implementation of the comprehensive rare disease plan and accompanying programs. At this time, it is not possible to estimate any cost savings on the part of individuals or the commonwealth once the comprehensive rare disease plan is implemented.

Conclusion

In conclusion, the Massachusetts Rare Disease Advisory Council (RDAC) was convened 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 a rare disease in the Commonwealth.

To date, the council has focused on establishing a foundation for its future work, including standard operating procedures, a mission statement, and strategic priorities. The council’s steering committee continues to provide direction and guidance for the work of the larger council. The council’s three workgroups have provided valuable data to support the council’s work.

* **Workgroup 1** has completed its prevalence study and produced its final report. (Appendix A) The prevalence report confirms that Massachusetts is in line with other predictions. The report estimates that a minimum of 244,369 to 432,882 people in Massachusetts have a rare disease.
* **Workgroups 2 and 3** continue to gather data and information through their survey tool. They are now focusing on a marketing plan to promote the survey with the goal of developing a profile of rare disease resources and social supports in Massachusetts as well as a profile of the expert individuals, community-based organizations, voluntary organizations, healthcare providers, and any other public or private organizations with an interest in rare diseases in Massachusetts.

Over the next year, the council looks forward to completing its data and information-gathering activities and performing the foundational analyses required to underpin the development of meaningful recommendations on improving the lives of people with rare diseases in the Commonwealth. The council plans to explore other opportunities to hear from the public and those impacted by a rare disease.

The council plans to identify and verify the social supports and services available for individuals and families with a rare disease in Massachusetts and identify the stakeholders interested in the rare disease community. As the council looks to complete its foundational work over the next year, it is mindful of its primary goal of developing meaningful recommendations to the governor, state legislature, and the Department of Public Health that will improve the lives of those impacted by a rare disease.

#  Appendix A

Prevalence of Rare Diseases in Massachusetts

(Approved by the Council on 9.21.23)

Executive Summary

Pursuant to the Massachusetts Rare Disease Advisory Council (RDAC) charter, we present this summary of the prevalence of rare diseases in Massachusetts. The purpose of this document is to provide the legislature with an estimate of the proportion of Massachusetts residents affected by a rare disease. To the best of our knowledge, no other RDAC in the country has prepared a similar report estimating rare disease prevalence within their state.

Despite the name, when taken together, rare diseases are not rare in society. Having a prevalence estimate is important so that the public and the legislators can have a sense of the magnitude of this problem and adequately prepare a response. Prevalence helps inform programs for family support, disease management, and diagnosis, facilitates access to care, guides policy decisions, helps in the allocation of healthcare resources, and informs clinical trial design.

We estimate that a minimum of 244,369 to 432,882 people in Massachusetts have a rare disease. However, rare disease patients are likely to be underdiagnosed and under-identified in healthcare systems, making it difficult to identify the true scope of their impact. Through policy and system changes, we can improve the lives of individuals and families living with rare diseases in Massachusetts.

Introduction

The members of the Massachusetts Rare Disease Advisory Council are pleased to present this summary of the prevalence of rare diseases in Massachusetts. The purpose of this document is to provide the legislature with an estimate of the proportion of Massachusetts residents affected by a rare disease. Having a prevalence estimate is important so that the public and the legislators can have a sense of the magnitude of this problem and adequately prepare a response. Through policy and system changes, we can improve the lives of individuals and families living with rare diseases in Massachusetts.

In the United States, a rare disease is defined as a condition having less than 200,000 cases. Rare diseases are medically diverse and can impact people across demographic characteristics. The exact number of people affected with rare diseases in Massachusetts is difficult to estimate for several reasons. The collective population of people with rare diseases represents a heterogeneous group of people affected by thousands if not tens of thousands, of rare diseases. Among these diseases, there may only be a few people affected in the Commonwealth for some of them. In aggregate, as we will summarize, there are many people affected by rare diseases. We estimate that between 244,369 to 432,882 people (between 3,500 to 6,200 per 100,000 people) in the Commonwealth of Massachusetts have a rare disease.

Methods for Prevalence Estimates

Our findings are based on evidence-based prevalence data from Orphanet and are extrapolated from the total population reported in the 2022 Massachusetts census.1 In the following sections, we provide our rationale for the assumption that rare disease prevalence in Massachusetts is similar to international data. If anything, this range is a conservative underestimate of the number of people in Massachusetts with a rare disease for at least several reasons. First, Orphanet does not include certain disease categories, such as rare cancers. Second, it does not fully account for underdiagnosis, misdiagnosis, and continual new disease discovery, which are all known challenges in rare disease identification. For these reasons, the National Organization of Rare Disorders (NORD) states that approximately 10% of people have a rare disease, which could raise the Massachusetts rare disease prevalence to close to 700,000 people.

To provide this estimate, we searched the peer-reviewed literature for national and global rare disease prevalence estimates. We relied on international data based on Orphanet, a portal for rare diseases that provides an inventory and classification of rare diseases. These Orphanet prevalence estimates are provided at a global or European level. Although there are likely to be differences in the population of MA and Europe, in terms of specific proportions of people from different ancestry backgrounds, the estimates are the most reliable systematic data points available.

A 2019 estimate using the Orphanet data estimated that 6.2% of the general population likely had a rare disease. 2 A 2020 global estimate using the same data, but a different methodology found between 3.5-5.9% of the population was estimated to have a rare disease. 3 This did not include infectious diseases or rare cancers. The National Cancer Institute estimates that 27% of all cancers are rare.4 According to the American Cancer Society, over 1.9 million new cancer cases are expected to be diagnosed in the U.S. in 2023.5 Both of the estimates were considered conservative by the authors, and the true population living with a rare disease is likely much larger. Our reported estimates were calculated using the range 3.5-6.2% of the population.

The United States has no centralized infrastructure to identify and track rare disease patients systematically. It is estimated that less than 5% of rare diseases have a specific International Classification of Disease (ICD-10) code, the identifier used in medical health and billing systems.6 Some rare diseases are included in newborn screening panels, have dedicated registries, or are tracked by patient organizations or medical centers with expertise in the condition. These methods can be used to collect more reliable estimates of the actual number of patients within that condition. However, patients affected by rare diseases are geographically dispersed, and may not participate in voluntary systems or connect with medical experts who are collecting information on a particular disease.

One challenge of determining disease prevalence specific to Massachusetts is that there may not be state-specific data on each rare disorder. To address this, we compared prevalence data that are available at both the national and Massachusetts level for some of the more frequent rare diseases that are accurately counted, such as by newborn screening, registries, or other reporting mechanisms. As outlined in the Appendix (Table 1), we found several examples of rare diseases where the Massachusetts prevalence matched the national prevalence. Concordance between national and Massachusetts prevalence rates strengthens our confidence in the accuracy of our estimated prevalence of rare diseases within the Commonwealth of Massachusetts.

General Challenges and Limitations

There are many challenges that make it difficult to accurately estimate the prevalence of rare diseases. Individual rare diseases are often poorly understood, may not have clear diagnostic criteria, and are often difficult to diagnose, with many patients waiting years to get an accurate diagnosis. It is often recommended that people with a rare disease undergo genetic testing, but many rare diseases cannot be diagnosed by genetic testing, or such testing may not be covered by insurance making it difficult for families to access. It is essential to understand the needs of undiagnosed rare disease patients when making healthcare policy recommendations. However, this patient population is even more challenging to identify and understand. It is equally difficult to determine the total number of rare diseases. New diseases are constantly being identified due to scientific breakthroughs and new genetic discoveries. Even within more common diseases, rare variants or subtypes can be identified.

Special Considerations for Massachusetts

It is possible that there is a higher percentage of rare disease patients in Massachusetts due to the breadth and quality of our healthcare facilities. Some evidence suggests that patients will relocate to be closer to specialists or to access needed healthcare, this could include individuals from other parts of the country or international patients engaging in either medical tourism or clinical trial participation. Additionally, the pharmaceutical and biotechnology industry in MA increases the potential for innovative discoveries and partnerships within the state that present unique opportunities for families and patient organizations, which may mean there are more families identified and diagnosed from within the state or who choose to relocate.

Importance of Prevalence

Prevalence is critical to provide context for the scope of a health problem. Understanding the number of people who have a particular health condition can impact the following:

* Family support: Programs can be designed to meet the needs of individuals living with a particular health problem, including establishing virtual or local support groups.
* Disease management and diagnosis: Medical professionals may be more likely to pursue a diagnosis if they understand how likely they will encounter a particular disease. Some medical professionals may not even consider an extremely rare diagnosis. Understanding how people with that diagnosis are treated and their prognosis helps inform healthcare decisions.
* Access to care: If there is a clearly defined population with a documented set of needs, it is easier to make the case for access to crucial healthcare needs, such as medical equipment, healthcare supplies, and access to specialists such as therapists and home nursing aides.
* Policy decisions: Once a population is defined, policymakers may identify ways to support the community further, including new studies to generate additional knowledge, additional programs to support patients and families, changes in the healthcare financing or delivery system, and protections within the insurance system.
* Allocation of healthcare resources: Budget considerations can ensure enough resources are dedicated to funding programs that provide families, healthcare providers, and the community with the necessary resources.
* Clinical trial design: Knowing the number of potential participants can help regulatory agencies decide the type of trial they are willing to approve and the number of people who need to participate to be considered a meaningful sample. It also helps companies anticipate the number of people who would benefit from drug development and which conditions to pursue further research.

Conclusion

Rare disease patients are likely to be under-diagnosed and under-identified (or mis-identified) in healthcare systems making it difficult to identify the true scope of the public health impact of rare diseases on our healthcare system and the vast unmet need for rare disease patients and families. There is an indication that the prevalence of rare diseases is growing, which is likely attributed to better diagnostics, more effective treatments, greater awareness of rare diseases, and new disease discoveries. This underscores the growing importance of establishing the necessary infrastructure to support rare disease patients and families and the healthcare providers that treat them. As the RDAC, we look forward to working with the key decision makers and the rare disease community in Massachusetts to improve the lives of all of those impacted by rare diseases in the Commonwealth.

Contributions

* Lead Author: Tai LS Pasquini, MPA, PhD
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Appendix

Figure 1: Comparison of Massachusetts Prevalence Estimates by Data Sources



Table 1: Select MA Rare Disease Specific Prevalence Estimates Compared to State Projections

|  |  |  |  |
| --- | --- | --- | --- |
| Disease | Estimated National Prevalence | Reported MA Prevalence | Comparison of Actual and Anticipated Prevalence |
| Spinal Muscular Atrophy (SMA)  | 1 in 10,000 live births7Estimated prevalence is between one and two per 100,000 people.  | .44 in 10,000 live birthsMA newborn screening study found 9 in 204,643 live births (January 2018-January 2021) 8 | Lower than expected |
| Sickle Cell Disease (SCD) | 45 in 100,000 138,923 patients (based on the birth cohort from 2005 to 2007)9 | 50 in 100,0003,261 patients (based on the birth cohort from 2005 to 2007)9  | Higher than expected |
| Hemophilia | 1.6 to 23.3 per 100,000 males10  | 11.7 – 14.4 in 100,000 males10 | Overlap in the mid-range |

Interpretation: Reliable Massachusetts state-specific rare disease estimates were difficult to identify for individual diagnoses. We found that for some diseases, MA data were reasonably close to national estimates. For others, there are challenges (as outlined above) in obtaining accurate direct counts or estimates. Nonetheless, on the whole, our working group felt that general population level estimates are adequate to project the total number of rare disease patients in MA.

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#  Appendix B

Rare Disease Advisory Council (RDAC) Survey

(ENGLISH version)

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 health care system that puts patients first ([Section 26 of Chapter 260 of the Acts of 2020](https://malegislature.gov/Laws/SessionLaws/Acts/2020/Chapter260)).

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 a rare disease 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 with the goal of improving 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 report of the council. WE NEED YOUR HELP. Please forward to anyone you think would be helpful in completing information.

Please note: All information given is confidential and your participation is voluntary, but 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, 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 non-profit?

○ Private
○ Non-profit
○ 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.”

#  Appendix C

**Massachusetts Rare Disease Advisory Council**

**Virtual Town Hall**

**Saturday, October 28, 2023**

**Special Report**

**Executive Summary**

The Massachusetts Rare Disease Advisory Council (RDAC) held its first virtual town hall on Saturday, October 28, 2023, to listen and learn from the public and the rare disease community. The meeting was open to the public, and stakeholders in the rare disease community were invited to provide testimony. The purpose of the town hall was to gather information about the availability of resources needed by those living with a rare disease in Massachusetts.

The central theme of the rare disease town hall testimonies focused on challenges in accessing therapies and social supports. Multiple testimonies referenced a lack of insurance coverage for costly treatments for people with rare diseases. Lack of access to home care was also cited as a major challenge to the quality of life for people with rare diseases.

Other themes heard during the oral and written testimony included:

1. Insurance coverage for rare disease care

* Challenging pre-authorization process
* Lack of coverage of drugs and treatments
* No coverage to see out-of-network providers/specialists

2. Need for home care

* Challenges to finding quality, reliable home care staff
* Poor pay for care attendants resulting in high turnover and unreliable services

3. Difficulty using transportation supports

* Unreliable services (“the ride”)
* Unable to access rides like Uber and Lyft

4. Utility of telehealth

* Need improved access to telehealth services
* Desire for coverage of care provided by telehealth

These testimonies offered at the town hall will help the RDAC to make meaningful recommendations to the governor, the legislature, and the Department of Public Health about how to improve the lives of people with a rare disease living in Massachusetts.

**Oral Testimony**

*As a member of the Rare Disease Advisory Council member, I am sharing my knowledge and experience as a mother and caregiver to my three children who live with a rare disease. I have two sons with Deschenes Muscular Dystrophy (DMD) and one son with Primary Immune Deficiency (PID). I wanted to share what is going well so we don’t just talk about what is going wrong. My oldest son, Austin, is on the call and will speak shortly, but I wanted to talk about a wonderful resource that we just recently learned about. It’s the Complex Care Team at Massachusetts General Hospital. I learned about this program through this council, and it has been a great resource for us. Austin is a frequent flyer to Mass General Hospital, and this Complex Care Team has helped us navigate some of the challenges associated with regular admissions to the hospitals.*

*One of the biggest challenges we have faced is access to innovative medications and treatments. Although Massachusetts has a strong biotech presence, access to some of its innovative drugs is limited. For example, my two older sons are wheelchair users, and the only medications approved for DMD are not available for wheelchair users. We have been trying to get access to these innovative drugs and treatments for almost ten years without success.*

*Another big challenge for me as a caregiver is home care. My primary goal has been to keep my sons home, and home care is a critical piece of that. As a single mom who also has a full-time job, home care becomes one of our primary needs. Finding quality and reliable home care has been a real challenge.*

*Lastly, I want to say that I’m happy to be living in Massachusetts. It has a lot to offer as far as medical care, and I know we are very lucky, but improving access to critical resources and social supports is necessary to improve the lives of those living with a rare disease in Massachusetts.*

Jenn McNary, Saugus

*I am Austin, and I am living with a rare disease. I am twenty-four years old, and the most important thing to me is my independence. In order to have that independence, I need to rely on services like “the ride.” I have a lot of medical appointments, and “the ride” is not always reliable. It doesn’t always get me to my appointments on time. There often isn’t anyone available by phone to verify my pick-up time or even confirm that my ride is scheduled. This adds a lot of stress.*

*I rely on people almost 24 hours a day to ensure my independence. Personal Care Attendants (PCAs) are a critical part of my care team. I would like to say that PCAs don’t get paid much, and they often don’t stay at the job long because of the pay. This makes it hard for me. When I don’t have caregivers, I have to rely on my mom and my 16-year-old brother for my care. My brother has his own rare disease and should not have to care for me. He has his own life, and we are worried now because he is looking at going to college and, therefore, won’t be available for backup care. I now need a BiPAP (a non-invasive ventilator) at night. I’m still getting used to it. I had a near-death experience due to RSV, so I now need a BiPAP at night. I think I should be able to rely on nursing and home care to help me, especially at night, but insurance won’t approve it. It really bothers me that people think that my mom should take care of me. It’s not fair. I don’t understand why it is expected that my family takes care of me when I’m an adult. I hope people realize that nursing is keeping me out of the hospital, and I don’t understand why I can’t get approved for the nursing hours I need to stay healthy and active and out of the hospital. People seem to think I should be in a facility that is a lot more expensive than living at home, besides the fact that other people may be sicker and need that hospital bed more than me.*

Austin LeClaire, Saugus

*I’m Criss Quigley, I’m here representing people with rare craniofacial disorders. They are dentinogenesis imperfecta, amelogenesis Imperfecta, and ectodermal dysplasia. We are a small group, and our primary challenge is insurance coverage for treatment. To give some background about this birth defect, it is a birth defect that affects the teeth, soft tissues of the mouth, and the jaw. Teeth become nonfunctional over time. Kids have trouble eating and often need a liquid diet. The social stigma and isolation that occurs is very damaging. It also affects one’s ability to get a job and be productive. Most treatments are not covered by insurance. Parents often learn that the treatment will cost them thousands of dollars. Research shows that treatments for these conditions can cost as much as half a million dollars. Parents go to extreme measures to find the funds to pay for these treatments.*

*Fifteen years ago, when my daughter’s out-of-pocket expenses exceeded $150,000, I decided to try to do something. I found another family who had a child with a similar condition, and we worked with our legislator to get a bill written that would ensure insurance coverage for these conditions. A similar bill was presented at the same time for people with cleft lip and palate. The bills traveled together from 2009 to 2013, when the bill for cleft lip and palate passed but ours did not. The bill has been refiled seven times and still has not passed. Our families have met with many advocacy groups, hospitals, and dental schools and have testified in front of the legislature many times. Our children deserve treatment as much as others. We are a small group, and we hope that the Rare Disease Advisory Council can help us.*

Criss Quigley, Springfield

*My name is Jillian, and I am the mother of a little girl who has a rare disease. She has OCNDS or Okur-Chung neurodevelopmental syndrome. She presents with epilepsy-type symptoms, and her condition is considered ultra-rare. Because of that, most doctors, nurses, teachers, and caregivers do know about her disease. I find that I have to make up education sheets to help people understand her condition. I believe that one of the biggest problems of having a rare disease is education. I feel very lucky that we have access to services like early intervention, Physical therapy, occupational therapy, etc, but it’s really hard explaining her condition to everyone.*

*It is heartbreaking to hear that kids are not getting the treatments they need due to costs, but I know it’s a reality. All kids should get access to the medications and treatments they need without worrying about costs.*

Jillian Kavanaugh, South Hamilton

*My name is Tai. I am a member of the RDAC, and I became a member to contribute as a researcher, but recently, my mom has shown signs of cognitive decline. With a family history of a rare neurocognitive condition, we became concerned. My mom recently moved from Connecticut to Massachusetts to be close to us. She had to change all of her doctors. We have had a very difficult time finding specialists for her to see. The wait times to get into a neurologist are really long. Medicare tried to find a plan that would allow her to go back to Connecticut to see some of her doctors in the meantime, but the plan they picked was not a good fit. We also realize that doctors don’t know about this rare condition. She needs to see a specialist, but it’s almost impossible to find a specialist near us. We are not sure if my mom has the condition at this point, and we don’t know when we may need to travel to find a doctor who can diagnose and treat her. It’s really hard to know when to do this. It's crazy that insurance is preventing us from accessing the right doctor for her.*

*As a supporter of a single-payer healthcare system, I know that one of the biggest complaints is long wait times, but we have long wait times now. There needs to be a better system. People should be able to get the care they need without worrying about networks, formularies, and other restrictions that are insurance-driven. Thank you.*

Tai Pasquini , East Longmeadow

*I am a mom and caregiver of a little boy with three rare diseases. One of the challenges I have experienced is the difficulty of getting a diagnosis. A lot of times, you can’t access services if you don’t have a diagnosis. We need to fix that. It sometimes takes years to get to a diagnosis, yet the child and family need help while trying to figure out the diagnosis.*

*The other topic I’d like to talk about is home care. As stated earlier, home care is critical. The PCA program sounds great in theory, but it’s broken. PCAs are great when you find a good one, but at $18/hour with no benefits, there aren’t a lot of people wanting to do this work. Personally, I don’t know why a family can’t be paid as a PCA. Currently, my 80-year-old mom is providing these services for my son, but it’s really hard. Many families can’t work because they need to stay home to care for their child. If they could be paid as a PCA, it would help.*

Ali Mahady, Westford

**Question and Answer Session**

Question: Coverage for treatment and equipment continues to be a challenge for many with a rare disease. Is it MassHealth or private insurance?

*My daughter currently needs treatment, and both dental and health insurance have denied her to get this treatment which will cost over $100,000. Most of the families we talk to have private insurance, but I can honestly say that I don’t know one family that has got insurance coverage for their treatment. To answer the question, I don’t know if it’s a MassHealth or a general insurance issue.*

Criss Quigley, Springfield

Question: If you could remake “the ride” what would it look like? Have you ever used Uber, if so, does that work?

*First, I think that the ride should be free. Second, there should be some dependability. When I have an appointment, I need to get there on time. Also, sometimes, the drivers will just leave us, even if we missed our appointment. Unfortunately, accessible Uber rides are hard to find.*

Austin LeClaire, Saugus

Question: We heard that specialists are hard to find. Do you know if there is a way to find specialists in other areas?

*My daughter’s specialist is at Boston Children’s Hospital, so we are very lucky. Although we are close to her specialist, there are other healthcare providers we need to see, and they know nothing about her diagnosis. Bringing a fact sheet whenever we go to a new provider has been the best solution for us.*

Jillian Kavanaugh, South Hamilton

Question: Can you speak to the PCA program? What would you like to be improved about it?

*The first thing would be to pay. PCAs are getting roughly minimum wage. The work is hard. People need to be responsible. It’s not babysitting. My son needs to be spoon-fed close to 15 times a day. He is blind, so he needs ongoing assistance. PCAs should get benefits. Other states allow parents or other family members to be paid as a PCA. It’s also hard to get the number of hours you really need. Also, nursing hours are important. MassHealth seems to be cutting nursing hours when they are clearly needed.*

Ali Mahady, Westford

Question: How does the PCA program work?

*The PCA program is a state program of MassHealth. There is a fiscal agent called Tempus who pays the bills, but it is up to me to interview, hire, fire, and manage the schedule and timesheets of the PCA. The family is also responsible for training the PCAs. It is often a full-time job, just managing the PCAs and the schedules. There are many barriers to the coordination of care when using PCAs.*

*The home care situation is a significant problem for those living with a rare disease in Massachusetts. The PCA program is broken and needs revisions. We know there are many issues related to home care to address, and we hope that the RDAC can tackle some of them.*

Jenn McNary, Saugus

*I know there is a PCA union, but I’m not sure how much they have been able to change for PCAs in Massachusetts. I think the pay has gone up a little but not enough to make a difference. MassHealth breaks down to minutes you get for the year. If your child needs to have a diaper changed and be fed, the PCA isn’t going to come just for an hour. MassHealth needs to relook how they calculate the time that a child needs. My friend, who is in another state, has to calculate how much time it takes to feed her child. As we all know, feeding a child one day may take 20 minutes, and feeding on another day could take 40 minutes. I believe that MassHealth is going to start requiring PCAs to clock in and clock out when they come to provide services.*

Ali Mahady, Westford

**Written Testimony**

***Please provide testimony on the most important healthcare resources and social supports that currently exist and how you found them to be valuable (up to 500 words).***

*I am responding as a friend of several Massachusetts ALS patients. Two of them have died during the several years I have been involved in ALS advocacy. ALS is a devastating diagnosis. Families impacted by ALS deserve and require extensive support. We are fortunate in the greater Boston area to have support from CCALS and ALSA, along with drug research by TDI and area hospitals. However, many regions in MA lack these resources. For patients with ALS, access to trained home health care aides and adaptive equipment should be available statewide.*

Jennifer Wright, Wakefield

*I am specifically responding with second-hand (as an advocate, not a patient or caregiver) to advocate for change in the treatment/care of patients with ALS. I am a member of the Massachusetts ALS Champions (www.palsmac.org), a non-affiliated group of local advocates for change in the ALS landscape. My perspective is shaped by watching Becky Mourey and Cathy Nally, and their families, fight hard and struggle through the challenges of ALS. I understand that dedicated ALS clinics provide top-notch care but are not always accessible/available to patients from outside the areas where these clinics are located. Organizations such as CCALS and the ALS Association (ALSA) provide fantastic support and care services to families dealing with ALS, such as equipment (power wheelchairs, vans, communication tools such as Tobii, etc.). ALS organizations, including CCALS, ALS One, I Am ALS, and ALSA provide networks and social supports.*

Barbara Kipp, Bedford

*We are primary care pediatricians who have a combined 60 years of caring for infants to young adults with complex and chronic illnesses here in the Commonwealth. Many of our patients have so-called rare diseases or congenital issues or conditions that have yet to be named or fully understood. Like many in this forum, their family experience is made more challenging by the lack of reliable sources of information and the sea of misinformation and questionable assertions online, including social media. We have shared the learned lessons of what works better and, at times, even best. We learned this early: parents are often the true experts in the particularities of their child’s health and offer critical insights to make optimal shared plans of care. In turn, a family and care team must learn to work collaboratively to seek the best evidence and weigh the risks and benefits of treatments. While the provider and care team relationship may be important in promoting wellness and preventing illness, it is by no means the only source of support for a family. Other parents, peer mentors, health coaches, and online networks can offer the benefits of community and shared experience. From referrals made by palliative care providers to online platforms like the Courageous Parents Network and more, caretakers may develop friendships and associations that provide a crucial and rare layer of support and perspective. We cannot say enough about the excellent work of organizations in the Commonwealth like MassFamily Voices or from the Federation of Children with Special Needs who can help families address whatever their evolving needs may be, including their physical, emotional, financial and spiritual health over time. While in the past few decades, there have been great advancements in the care of children with disabilities allowing them to live outside the hospital and for longer periods of time, supports for families have not been adequate nor equitable. Longer survival times have not necessarily resulted in a better quality of life for patients and their families. Families are burdened by hidden costs and challenges to even health-literate patients and families, including service gaps and the opportunity costs faced by caretakers foregoing employment opportunities to administer care for their child and/or to navigate health systems. As a result, these children may now be outliving their parents and will very likely live beyond their ability to be cared for by them. We now have a moral obligation to patients, their families, and providers to develop infrastructure to support in-home and alternative settings for this growing population. We want to use our testimony to draw attention to some of the great resources like MassFamily Voices or from the Federation of Children with Special Needs that exist for families of children with rare diseases and raise awareness of our collective need to better support the families impacted by what can be devastating or costly conditions that affect entire households. We also want to advocate for sustainable strategies for aging parents to support their families for the years to come.*

Jack Maypole, Waban

*MA offers good Medicare options. We also have excellent medical institutions that not only provide excellent care but also do incredible research in the rare disease space. The life science industry here is second to none.*

Steven Kowalski, Boston

*In October 2020, my sister and best friend Cathy Nally was diagnosed with ALS. Cathy faced ALS with the same love of life, determination, courage, and energy that marked her impactful 62 years on this earth. Widowed in 2001, Cathy was fiercely independent and raised two amazing children on her own. That independence served her well as she battled ALS; she tried to identify and address her ever-increasing needs by herself. It soon became clear to those of us who loved her that such an approach was not sustainable. Living in the north end of Boston, she was blessed to be so close to MGH and the Healey Center, where she received much of her world-class care and support. Most other ALS patients in MA are not so fortunate. But as ALS progressed, Cathy and I learned that she couldn’t do it by herself. A dear friend who lost his brother to ALS connected me with CCALS based here in Massachusetts. They were an incredible resource to my sister and our family, and I have recommended their services and programs to other families suddenly facing the uncertainty that comes with an ALS diagnosis.*

Ed Hurley, West Yarmouth

*The most important healthcare resources in our 30-year experience as an active organization supporting our rare disease community are proactive, well-organized, and well-funded nonprofits that are formed to work closely with healthcare providers and other stakeholders to assess community member's needs and do all it can to fulfill them, socially, financially and otherwise. ‘It takes a village’, as they say, and nothing could be more true than in the rare disease community. With rare disorders, it’s important for the community (patients, caregivers, care providers, industry partners, governmental agencies, and other nonprofits) to come together, stay connected, share information, support each other, and advocate for one another. Coping with a rare disorder is hard enough in and of itself. Remaining positive, managing the disorder on a daily basis, and continually advocating for oneself/family while planning for the future is even harder. Meeting people “where they are” at and helping them improve their health care is essential to ensure a healthy community. NECPAD often receives referrals from local clinics or individuals who reach out to us directly. Some just need someone to talk to about their diagnosis (not from a clinical perspective, but practical) and what the next step is in terms of how, in practice, they will address the diagnosis and ongoing care of an individual; others need some help financially to afford deductibles, copays or medically prescribed items not covered by insurance, others need help fighting for, and affording, insurance coverage (this is huge), others need help purchasing essential items to help them manage their care such as an extra refrigerator to store medically prescribed nutrition or transportation costs to clinic visits, others need legal advice, others need assistance with other basic life needs that are related to their disorder, and yet others just want to be connected to a community which “gets it” and can be there to lend advice, support and frankly, some respite! All are equally important and essential to help live, as well as possible, with a rare disorder. NECPAD strives to achieve this and inspires other stakeholders to work together to do the same. Thank you for the opportunity to comment and please do not hesitate to reach out to me so that we may continue this important discussion and work together to define and implement ways to improve the lives of those living with rare disorders in the Commonwealth of Massachusetts (and beyond) as Massachusetts has always been a leader in this regard (e.g. first state to implement Newborn Screening Program (NBS) and leader in healthcare and biotech industry)!*

Denise Queally, Chilmark

***Please provide testimony on the healthcare resources and social supports that are lacking or are difficult to access and how they would improve the lives of people with rare diseases (or their families if they did exist) (up to 500 words).***

*As mentioned above, access to support for families coping with ALS varies widely in MA. Non-profits can help with adaptive equipment to some extent. The biggest problem is a lack of home healthcare aides specifically trained to assist ALS patients. Care often falls to family members, with little or no respite. And even if qualified help were available, most families do not have the financial means to pay for such support. Patients with ALS should be provided with trained, well-paid home health support funded by private or government insurance.*

Jennifer Wright, Wakefield

*Despite some of the great resources mentioned above, it seems that ongoing care for people living with ALS (PALS) still falls mostly on family members. PALS should not have to rely on family members (who are also trying to take care of other family members and support themselves) for their care. While insurance (including Medicare) sometimes covers nursing/home health care, many/most of these workers are not skilled or trained in the needs of PALS, which includes dealing with respirators, feeding tubes, electronic communication equipment, patient lifts, etc. Some of these specialized skills apply to patients with other diseases too. Concerted efforts to increase the number of providers with these skills is needed, including better pay for the workers and specialized training (perhaps even “certifications” in specific skills). I understand that funding is available for this, but it has not been applied in a disciplined and focused way. I think the government should partner with the aforementioned ALS organizations to make this happen and should be held accountable. For people who don’t live near established ALS clinics, programs need to be developed/expanded to enable PALS and their families to participate in clinical trials and be treated by world-class clinicians. This will necessitate funding for care (including insurance coverage), transportation, and temporary housing arrangements. This greater access will help ensure that the care (and clinical research) surrounding ALS is representative of both the full disease population and the diversity of our country (including ethnic and socio-economic diversity).*

Barbara Kipp, Bedford

*As an affected constituent and parent of an affected child with rare dental disorders, ensuring access to medically necessary treatment for individuals with these conditions is an essential step toward achieving equitable healthcare, and I would greatly appreciate your support and advocacy. I am 49 years old and have seven dental implants that my parents were able to help pay for over the years. I am extremely fortunate to have parents who worked hard and were able to help pay the large dental bills for me. My son Jeremy (see pictures below) is 20 and has just completed his first major procedure. Although he does not have implants yet, what you see in the picture will cost my family $27,000 dollars, and this is just for the front teeth. He is a junior at Boston University (he has siblings at UMASS Dartmouth, a senior in high school, and a freshman in high school), so we will be helping to pay college tuition bills for years to come. This extra dental bill is a tremendous burden for our family. Any help getting this bill passed is greatly appreciated. Children and adults with rare craniofacial disorders affecting the mouth, jaw, and teeth experience pain, difficulty with eating and speech, bone loss, infection, jaw dysfunction, and psycho-social difficulties due to their facial differences. Although physicians categorize treatment as medically necessary, health insurance companies currently use loopholes to deny coverage to this group. These insurance denials result in individuals or families facing catastrophic out-of-pocket medical expenses for treatment, upwards of hundreds of thousands of dollars. Patients with cleft lip and palate are currently covered for dental treatment in Massachusetts, but those with more rare craniofacial disorders are not. This is not only unjust but also counterproductive to the overall well-being of our community. By providing insurance coverage for rare dental disorders, we can ensure that affected individuals receive timely and appropriate treatment without the heavy or impossible financial burden that often accompanies these conditions. This legislation would not only alleviate suffering but also promote better oral health outcomes, reduce the long-term healthcare costs associated with untreated dental conditions, and ultimately enhance the quality of life for those affected by these rare disorders. According to the most recent CHIA study, the cost to the insured population is pennies per month.*

Gregory Gay, Taunton

*Lack of insurance coverage for treatment for rare craniofacial disorder: My 12-year-old daughter, Elayna Melendez, born July 2, 2011, has been diagnosed with Amelogenesis Imperfecta (AI). AI is a condition that causes teeth to be unusually small, discolored, pitted or grooved, and prone to rapid wear and breakage. As of right now, I have already started to pay out of pocket for crowns for her teeth in the back of her palate that have been through rapid breakage. I am emotionally stressed and worried about how much I would have to pay in the future for a new set of teeth for my daughter as well as the maintenance of costs. Since Kindergarten, Elayna has been stressed, sad, worried, and upset, and I can name many more emotions that she feels about not having normal teeth. When people without a teeth disorder hear the word SMILE, it’s a positive thing. But for Elayna, it’s not. Smiling with AI teeth has made Elayna an easy target to get bullied. As a mother, I try my best to stay strong and tell her one day we will fix her teeth, and each time I tell her that, I wonder if it will be fixed or if I will have the funds to fix her teeth or smile when that day comes. My daughter bed wets still, and I believe her stress with her teeth may play a big role in it. My child isn’t allowed to eat certain things because of her condition; her teeth are so sensitive that sometimes she tells me she prefers not to eat. Parents normally worry or must plan to save for college for their children’s future. For parents with children with AI, it’s stressful thinking about saving for new teeth for their child. Or if that’s even possible. I’ve learned and heard of people paying $100,000 for new teeth, and that’s not including the maintenance. I hope this testimony gives you a realistic idea and understanding of what it’s like to have AI and also to be a parent to a child who has AI and the uncertainty of how much it’ll cost for treatment, the worrisomeness of just going to a dental appointment and just helping your child handle bullies, insecurities, and much more.*

Jasside Carvalho, Agawam

*I have been receiving treatment for my rare craniofacial condition, dentinogenesis imperfecta (DI), since I was two. People with DI have abnormally formed teeth — small, with tiny roots, grayish-brown, with no hard-white enamel so that they wear down or chip away without treatment. The condition is very painful and patients lose teeth easily. Patients may have difficulty eating or speaking, and the jaw may become wasted or deformed. In some cases, the jaw may even lock and not be able to open. These are serious, debilitating disabilities and treatment is complex, requiring a team approach. Without early treatment, my teeth would have worn down to gum level from normal chewing. I was fortunate that my parents were able to pay for a full mouth reconstruction at 16, and since then, I’ve continued to require treatment for infections, an open bite, broken teeth, loose and broken prosthetics, and jaw pain. The cost has been over $150,000 so far, almost all out of pocket, because Massachusetts law contains a loophole where insurers can deny coverage for birth defects that affect the teeth, even though the patient’s nutrition, speech, psychological well-being and quality of life are impacted by the disorder. Although the cost has been a huge burden on my family, I am fortunate compared to many who can’t afford the type of treatment I’ve had. I’m 35 now and just beginning another full mouth reconstruction, which will mean more oral surgery, bone and tissue grafting, implants, and all new crowns. It’s not uncommon for people with DI to need ongoing treatment throughout their life. The cost for this phase of treatment is estimated at least $110,000, and both my medical and dental insurance have already denied coverage. I delayed treatment much longer than was recommended, hoping that legislation mandating insurance coverage for these rare conditions would pass, and meanwhile dealing with broken crowns, an uncomfortable temporary denture, an increasingly limited diet, the risk of bone loss, and anxiety about the cost of treatment and my future. I am part of the Massachusetts advocacy group that has been working to pass legislation to mandate insurance coverage for patients with these rare disorders for the past 15 years. Since 2012, patients with cleft lip and palate have had mandated insurance coverage for dental treatment related to their craniofacial birth defect, but rare disease patients were not included in that mandate, so families like mine continue to face hundreds of thousands of dollars in out-of-pocket costs. Health insurance should be inclusive and not discriminate based on diagnosis or the prevalence of a condition. Mandating insurance coverage for these rare disorders would ensure that children and adults with these rare disorders have equal access to necessary medical care. Society as a whole benefits when all its members have improved health outcomes and quality of life.*

Ann Quigley, Chilmark

My name is Karen Tenner. I was diagnosed with Amelogenesis imperfecta, a condition in which enamel or the hard outer layer of teeth does not form, leading to soft misshapen yellow teeth. *As a result of this, I have dealt with countless psychological, emotional, functional, and financial challenges. I am now a practicing primary care physician in the Commonwealth and know firsthand the impact poor dentistry has on many aspects of my patient's health. Early on, my teeth were brittle and brown, crumbly. I was lucky my family sought out consultation from experts across the world, and I was diagnosed with AI at age 2. As an elementary school child, I still remember the taunts, the questions, “Don’t you brush your teeth,” and the discomfort and pain of eating that came with having enamel-less teeth. As a teenager, I spent a summer recovering from a bone graft to prepare my mouth for essentially a full set of implants. During my first two years of high school, before this drastic intervention, I was missing 2/3 of my expected adult teeth. I remember hiding my smile, ashamed of the gaping holes in my mouth. I am one of the lucky ones. I speak before you after almost every single adult tooth has been replaced. I have almost an entire set of dental implants. While my bone graft was partially covered by my medical insurance, there was essentially no other coverage. Over the course of my childhood, my parents estimate that they spent over 250,000 dollars on my dental work. Even with all of these interventions, my problems continue today – in the last three years, I have had three teeth pulled due to AI-related complications and implants placed. Now as an adult with excellent dental coverage, this work has still cost thousands of dollars out of pocket. This work is not just cosmetic. Teeth are essential for speech, sustenance, and our ability to function in the world. I am almost certain that had I not been born into a family that was able to afford this care out of pocket, I would not be the same contributing member of society I am today. As a patient and a physician --- I ask for your support in this essential bill to guarantee coverage so that all patients, regardless of their financial status, can get the care they so badly need and deserve as constituents in the commonwealth with these challenging craniofacial diseases.*

Karen Tenner, Arlington

*Increasing Access to Telehealth Services: Virtual care improves patient access to high-quality care well beyond the COVID-19 pandemic. More communities than ever have experienced telehealth’s powerful impact in bridging gaps in care, especially underserved and rural communities that historically have had limited access to specialty care can now see top specialists due to expanded access to telehealth. Banning the Use of Quality-Adjusted Life Years (QALYs) and Equal Value Life Year Gained (evLYG): QALY and similar cost-effectiveness metrics are discriminatory towards those with disabilities and rare diseases like ALS. State governments should ban the use of discriminatory QALY and evLYG metrics when determining coverage of existing and new ALS therapies. Eliminating or Decreasing Durable Medical Equipment (DME) Taxes: In general, in the U.S., tangible personal property (i.e., items you can see, touch, and interact with) is considered taxable when sold at retail. However, states can determine what is and is not taxable within their borders, and many states make exceptions for necessities like medical equipment. Taxes on DME unnecessarily increase the financial burden of people living with ALS. Guaranteeing All Copays Count Towards the Cost of Drugs (Banning Copay Accumulator Adjustment Programs): Many insurers have disallowed copay assistance (also known as copay savings program, copay coupon, or a copay card) from counting towards a patient's annual deductible or out-of-pocket maximum. As a result, people living with ALS can face unexpected costs of thousands of dollars to get the medicines they need. Reforming the Prior Authorization Process: As a cost-control measure, health insurance plans often require physicians and other health care professionals to obtain advanced approval from a health plan before a specific service or treatment is delivered to the patient to qualify for payment coverage. Prior authorization delays treatment and negatively impacts health outcomes. Combating the use of "Fail First" Policies: Often, insurers use fail first policies (also known as Step Therapy) that require patients to fail on the insurer's preferred drug(s) before a patient can take the drug originally prescribed. People living with ALS should have barrier-free access to the medications as prescribed by their providers.*

Steven Kowalski, Boston

*I was honored to help to provide care and support for my sister during her battle with ALS. My experience highlighted- something that was reinforced as I met other families facing similar challenges -that the care for individuals living with ALS falls primarily on immediate family members. There are limited specialized medical resources available for families and an acute shortage of trained individuals to support the medical and emotional needs of ALS patients. What CCALS does every day for families is amazing and very much needed. But as a lifelong resident of the Commonwealth, I would like to see our state take more of a leadership role in working with ALS-focused organizations and the medical community to provide needed programs and medical support for families facing the emotional and financial burdens of an ALS diagnosis. I promised my sister, before she passed in August 2022, that I would do everything I could to continue her powerful legacy of being an advocate and work to increase ALS awareness with the hope of positively impacting the lives of current and future ALS patients. I stand ready to help the future efforts of RDAC and others in MA to shine a bright light on the needs of an ever-growing number of families in my home state. Thank you very much for providing the opportunity for me to share my personal experiences with ALS*

Ed Hurley, West Yarmouth

*As a pediatrician practicing for over 35 years in Western Massachusetts and for the past 25 years at Baystate Children’s Hospital in Springfield, I would like to wholeheartedly support the statement of my colleague and Chair of the Massachusetts Chapter of the American Academy of Pediatrics Committee on Children with Disability, Dr. Jack Maypole. Great advances have been made for children with rare diseases since I started practice in the 1980s; unfortunately, there has been a progressive contraction of supports available to families. Fortunately, the Massachusetts Department of Public Health Division for Children & Youth with Special Health Needs (DCYSHN), Community Case Management, Family Voices, and the Federation of Children With Special Needs have bravely provided lifelines to our families. Our families of children with rare diseases are in crisis. There is a current shortage of many services in Western Massachusetts; these include Home nursing shortage, Durable Medical Equipment shortage, Limited availability of pediatric specialists with long wait times, Pediatrician shortage, Nursing shortage, Pediatric hospital bed shortage, Lack of respite services outside of eastern MA Complex prior authorization process of life-saving medication and treatments Medication cost – specifically the new biological agent's shortages of generic medication Limited supply of influenza and covid vaccines Hopefully as Mass Cares for Kids starts this January, we will be able to help comfort families, but until funding is devoted to this population we will continue to deprive families of the resources they need to thrive. This week the American Academy of Pediatrics announced a major new policy statement on Medicaid and CHIP, calling for bold changes in the program and a vision for what Medicaid and CHIP could be. I urge you to look at this resource, “Medicaid and the Children’s Health Insurance Program: Optimization to Promote Equity in Child and Young Adult Health,” in the November issue of Pediatrics as a blueprint for positive change.*

Matthew Sadof, Springfield

*For our nonprofit, we are all volunteers, and it is very difficult to dedicate the time and resources needed to properly perform outreach and address all the needs that are out there in our rare community. It would be great if we could actually have enough funds to pay for staff, and also to be able to do things like raising awareness and funds, marketing, communications, interactive website, survey capabilities, etc., so we can reach and support people! Also, in the PKU and Allied Disorder world, what is lacking is getting on the same page to share information, discuss our needs with one another and other rare disease communities and stakeholders, and identify resources that are either needed or available to us, locally and nationally. What is also lacking is accessing adequate health care coverage for our essential treatments. In the PKU example, we are blessed to have the Newborn Screening Program (NBS), which successfully identifies our disorder at birth, we are in the minority of rare diseases (less than 5%) which has a treatment (we actually have a few treatments) available to us that will allow us to live as healthy a life as possible, but yet we do not have sufficient health care coverage to be able to afford and fully access our medically prescribed treatments. Thus, for PKU, we have actually lost HALF of our community to care, meaning half of our community of 16,500, about 8,000, are “lost” to care, meaning they are not under the care of a metabolic clinic being followed by professionals and are not living to their full potential (lack of executive functioning, not able to hold a job or maintain a healthy relationship, depression/mental illness and, much worse, may not survive). Here we have NBS treatments but no access to living our best lives! I do not know another rare disorder, or other non-rare disorder, that has lost HALF of its own to care. It is atrocious! We have been fighting down in DC since 2009 for proper healthcare coverage, and we still do not have it. It is truly frustrating, time-consuming, and counterproductive. What good are treatments and cures if we do not have people well enough in the clinic to hear about them, participate in clinical trials, and benefit from these advances? First things first! Legislators need to intervene to ensure we have proper private and public health care coverage so that the rare disease community can live its best life according to the medical advances we have out there (such as NBS and proven treatments). Thank you for the opportunity to comment, and please do not hesitate to reach out to me so that we may continue this important discussion and work together to define and implement ways to improve the lives of those living with rare disorders in the Commonwealth of Massachusetts (and beyond) as Massachusetts has always been a leader in this regard (e.g., first state to implement the Newborn Screening Program (NBS) and leader in healthcare and biotech industry)!*

Denise Queally, Chilmark

