**MASSACHUSETTS RARE DISEASE ADVISORY COUNCIL (RDAC)**

**FULL COUNCIL MEETING**

Thursday, November 17, 2022, 9:00-11:00 am

Approved by the council on January 19, 2023

**Welcome**

*Guest Participant Announcement by Dr. Tierney*

The public is welcome to join any of the RDAC meetings. However, only guests that are on the agenda will be allowed to speak during the meeting. For all guests on this call, could you please make sure that your video is turned off and your audio is on mute.

Thank you for your interest in learning more about the work of the rare disease advisory council.

**Roll Call to establish a quorum**

|  |  |  |
| --- | --- | --- |
|  | **Council Member** | **Present** |
| 1 | Charlotte M. Boney, M.D | - |
| 2 | Janis Creedon | - |
| 3 | Lisa Deck | x |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | x |
| 5 | Michael R. Green, M.D., Ph.D. | x |
| 6 | Julie D. Gortze, RN | x |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA | x |
| 8 | Lena Joseph, RN, CPN | arrived after roll call but before minutes |
| 9 | Representative Hannah Kane | Sofia Flionis present representing Rep Kane |
| 10 | Andrew A. Lane, MD, PhD | x |
| 11 | Representative Jay Livingstone | Audrey Hermann present representing Rep Livingstone |
| 12 | Jeff R. Livingstone, PhD | - |
| 13 | Diane Lucente, MS, LCGC | x |
| 14 | Alexsandra B. Mahady | x |
| 15 | Jenn McNary | x |
| 16 | David T. Miller, MD, PhD | x |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | x |
| 18 | Shivang Patel, Pharm.D. | x |
| 19 | Asma Rashid, MS, CGC | x |
| 20 | Michele Rhee, MBA, MPH | x |
| 21 | Robert E. Schultz, MBA | - |
| 22 | Celia Segel, MPP | x |
| 23 | Michael Sherman, MD | - |
| 24 | Glenda E. Thomas | x |
| 25 | Ryan Thompson, MD, MPH | x |
| 26 | Dylan Tierney, MD, MPH | x |
| 27 | Ann Wessel, MS, RD, LDN | x |
| 28 | Ross Zafonte, DO | - |

A quorum was established, and the meeting was called to order by **Dr. Tierney** at 9:05.

**Dr. Tierney** asked for a motion to approve minutes from the last full council meeting on 9.15.22

made a motion to vote on the minutes

seconded the motion

**ROLL CALL VOTE to Approve Minutes from 9.15.22**

|  |  |  |
| --- | --- | --- |
|  | **Council Member** | **Approve** |
| 1 | Charlotte M. Boney, M.D | - |
| 2 | Janis Creedon | - |
| 3 | Lisa Deck | X |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | x |
| 5 | Michael R. Green, M.D., Ph.D. | - |
| 6 | Julie D. Gortze, RN | X |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA | X |
| 8 | Lena Joseph, RN, CPN | x |
| 9 | Representative Hannah Kane | X |
| 10 | Andrew A. Lane, MD, PhD | x |
| 11 | Representative Jay Livingstone | X |
| 12 | Jeff R. Livingstone, PhD | - |
| 13 | Diane Lucente, MS, LCGC | X |
| 14 | Alexsandra B. Mahady | x |
| 15 | Jenn McNary | X |
| 16 | David T. Miller, MD, PhD | X |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | X |
| 18 | Shivang Patel, Pharm.D. | X |
| 19 | Asma Rashid, MS, CGC | X |
| 20 | Michele Rhee, MBA, MPH | X |
| 21 | Robert E. Schultz, MBA | - |
| 22 | Celia Segel, MPP | x |
| 23 | Michael Sherman, MD | - |
| 24 | Glenda E. Thomas | X |
| 25 | Ryan Thompson, MD, MPH | X |
| 26 | Dylan Tierney, MD, MPH | x |
| 27 | Ann Wessel, MS, RD, LDN | X |
| 28 | Ross Zafonte, DO | - |

**Dr. Tierney** introduced guest speaker and council member **Jenn McNary**

Navigation of care for adults living with rare health conditions continues to be a challenge even for families like Jenn’s who have the knowledge and resources to navigate the system. Care providers with expertise in these conditions are often hard to access, have long waitlists, or are located in buildings with inaccessible access. In addition, many non-specialists aren’t comfortable handling the needs of patients with rare diseases. Jenn is a caregiver to two adults living with Duchenne Muscular Dystrophy. She will talk to us today about her experiences navigating the difficulties of care access. She will share some examples and present some possible solutions.

**Jenn McNary** spoke passionately about the challenges of accessing health care, advocacy, and navigating the health care system. The emphasis was on how challenging it is for someone transitioning from pediatric care to adult care. Children with a rare disease are living longer, and the healthcare system is not prepared to provide care for these patients. She shared the story of her son, Austin, who is living with Duchenne Muscular Dystrophy. He has been hospitalized five times this year. He has lost weight due to his bowel issues and is challenged to get the care that is best for him. Primary Care Physicians are difficult to find as they do not want to work with someone with a rare disease. When health care needs happen, they are sent to the ER, where they experience long wait times (16 hours). Because Austin is now an adult, Jenn becomes his caregiver in the healthcare system, not his parent/guardian as she was during pediatric care. She has had difficulty getting in to see her son once he has been admitted (after hours). She is often not heard because she is seen as his advocate versus his parent. Hospitals are eager to discharge the patient before the goals for discharge are met. Patients are often sent home, and family members are expected to deliver the necessary complex medical care that is required. Patients are left to rely on personal care assistants or PCAs. They are paid at least $16 an hour, and they are often hard to find. Austin has faced a revolving door of untrained CNAs.

**Dr. Tierney** asked if there were any questions.

**Dr. Thompson** spoke about learning from one another. Patients are often left to learn about resources and even doctors who treat their condition through conventional and unconventional communication channels.

Jenn stated that Mass General is Austin’s preferred hospital because they have his records and can address his issues. If a patient with a rare disease goes to an ER that they don’t know, the ER is often unprepared to deal with their issues. Sometimes, non-emergency medical needs are addressed by being admitted to the hospital. Jenn usually has to ask Austin’s nurses to advocate for him when she is out of town.

**Julie Gortze** expressed her appreciation and how important advocacy is.

**Ali Mahady** spoke of her experience being referred to the ER and was then challenged to find PCA.

**Glenda Thomas** spoke about the challenges of finding a PCP that knows about your rare disease. She suggested developing a registry of doctors with rare disease experience.

**Michele Rhee** spoke about the challenges of finding a doctor or an advocate that can help. She said it is specifically difficult post-hospitalization if you do not have family nearby. She said there could also be legal challenges of a non-family member providing care.

**Dr. Tierney** asked Jenn what would be most helpful for her as Austin’s caregiver.

Jenn said that a focus on non-emergency care would be most helpful. Going to an ER and possible admission to the hospital often sets him back. Telehealth would undoubtedly be helpful. Having a telehealth virtual visit vs. waiting in the ER for 16 hours is helpful. Also, if hospitals could be better prepared to admit a patient upon arrival, it would be beneficial.

**Lena Joseph**stated that we should start a discharge plan when a patient is admitted.

**Lisa Deck**thanked Jenn for her tireless efforts to advocate for those with rare diseases. She hoped that Massachusetts could get into the fight with her.

**FULL COUNCIL DISCUSSION ON COUNCIL GOALS**

**Dr. Tierney** walked thru the slides. He added that the legislative language would help guide the work of the RDAC. The council reports to the legislature would include the council recommendations.

**Dr. Tierney** talked about how the goals should support the legislative charges given to the council. The council reports would be given to the legislature but would also be a resource for those living with rare diseases.

**Dr. Tierney** stated that the Steering Committee drafted goals to address the legislative charges. The committee recommended forming workgroups to do some of the initial foundational work. They also recommended that all council members be part of at least one workgroup. When these goals are accomplished, we can address new goals, such as the patient bill of rights.

**Tai Pasquini** spoke about her work and the challenges of forming estimates vs. surveys and registries. She asked if we knew what other RDACs have done to determine prevalence.

**Dr. Miller** focused on being practical. However, he also reinforced that determining the prevalence of rare diseases in MA would help support the RDAC’s other goals and recommendations.

**Michele Rhee** commented that the numbers could also support efforts to find new treatments.

**Ali Malhady** asked how determining prevalence would increase access to health care.

Dr. Tierney stated that determining prevalence was a legislative charge.

**Dr. Dwyer** stated that even a simple statement could be powerful.

**Tai Pasquini** talked about using literature reviews and hospital data as resources.

**Lena Joseph *asked*** if we knew how many rare diseases have a registry like cystic fibrosis.

**Tai Pasquini**stated that many diseases have registries, but participation varies.

**Dr. Thompson** suggested reaching out to MassHealth Long-Term Support and Services (LTSS) for goal 2.

**Lena Joseph**stated that we should survey advocacy information to ensure that we don’t duplicate our efforts.

**Tai Pasquini**stated that we should not use ICD codes as a data point. However, she stated that she is working on a project using ICD codes, and it’s very challenging. Needless to say, that may rare diseases don’t even have an ICD code.

**Michele Rhee**stated that she believes that biopharma does a lot of work on the prevalence and that we should reach out to them. MassBio could help.

**Tai Pasquini** stated that we should start with a literature review.

**Jenn McNary**stated that estimates of numbers could really help in future asks.

**Dr. Tierney** showed the slide with the organizational structure of the workgroups and requested that everyone volunteer to be on at least one workgroup. There is a lot of work to be done, and we need all to help. Each workgroup will also need a chair. Please let me know if you are interested in becoming a chair of one of the workgroups.

**Dr. Tierney** talked about how change may seem slow, but their work on the council will show the legislature the importance of the needed changes.

**Dr. Tierney** reviewed the council's priorities and the initial goals.

**Strategic Priority 1**

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

**GOAL:** Determine the prevalence of rare diseases in Massachusetts.

**Strategic Priority 2**

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

**GOAL:** Develop a profile of rare disease social supports and services in Massachusetts

**Strategic Priority 3**

***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.

Dr. Tierney asked for a vote to approve the initial goals.

**Tai Pasquini** motioned

**Celia Segel** seconded the motion.

**ROLL CALL VOTE** to approve these goals

|  |  |  |
| --- | --- | --- |
|  | **Council Member** | **Approve** |
| 1 | Charlotte M. Boney, M.D | - |
| 2 | Janis Creedon | - |
| 3 | Lisa Deck | X |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | x |
| 5 | Michael R. Green, M.D., Ph.D. | - |
| 6 | Julie D. Gortze, RN | X |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA | X |
| 8 | Lena Joseph, RN, CPN | x |
| 9 | Representative Hannah Kane | - |
| 10 | Andrew A. Lane, MD, PhD | X |
| 11 | Representative Jay Livingstone | X |
| 12 | Jeff R. Livingstone, PhD | - |
| 13 | Diane Lucente, MS, LCGC | X |
| 14 | Alexsandra B. Mahady | X |
| 15 | Jenn McNary | X |
| 16 | David T. Miller, MD, PhD | X |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | X |
| 18 | Shivang Patel, Pharm.D. | X |
| 19 | Asma Rashid, MS, CGC | x |
| 20 | Michele Rhee, MBA, MPH | x |
| 21 | Robert E. Schultz, MBA | - |
| 22 | Celia Segel, MPP | X |
| 23 | Michael Sherman, MD | - |
| 25 | Glenda E. Thomas | X |
| 26 | Ryan Thompson, MD, MPH | - |
| 27 | Dylan Tierney, MD, MPH | x |
| 28 | Ann Wessel, MS, RD, LDN | x |
| 29 | Ross Zafonte, DO | - |

Motion carries. Goals are established as state above.

**ROLL CALL VOTE** to establish subcommittee workgroups

**Dr. David Miller** made a motion to vote

**Tai Pasquini** seconded the motion

|  |  |  |
| --- | --- | --- |
|  | **Council Member** | **Approve** |
| 1 | Charlotte M. Boney, M.D | - |
| 2 | Janis Creedon | - |
| 3 | Lisa Deck | x |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | x |
| 5 | Michael R. Green, M.D., Ph.D. | - |
| 6 | Julie D. Gortze, RN | x |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA | x |
| 8 | Lena Joseph, RN, CPN | x |
| 9 | Representative Hannah Kane | - |
| 10 | Andrew A. Lane, MD, PhD | x |
| 11 | Representative Jay Livingstone | x |
| 12 | Jeff R. Livingstone, PhD | - |
| 13 | Diane Lucente, MS, LCGC | x |
| 14 | Alexsandra B. Mahady | x |
| 15 | Jenn McNary | X |
| 16 | David T. Miller, MD, PhD | X |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | X |
| 18 | Shivang Patel, Pharm.D. | X |
| 19 | Asma Rashid, MS, CGC | x |
| 20 | Michele Rhee, MBA, MPH | x |
| 21 | Robert E. Schultz, MBA | - |
| 22 | Celia Segel, MPP | x |
| 23 | Michael Sherman, MD | - |
| 24 | Glenda E. Thomas | x |
| 25 | Ryan Thompson, MD, MPH | - |
| 26 | Dylan Tierney, MD, MPH | x |
| 27 | Ann Wessel, MS, RD, LDN | x |
| 28 | Ross Zafonte, DO | - |

Dr. Tierney asked for volunteers to join at least one subcommittee workgroup. He asked people to write in the chat which workgroup they would like to join or to email him by 11/28.

He also asked everyone to indicate if they were interested in being a chair. We will need a chair for each workgroup. He also stated that if we do not hear from anyone by 11/28, we would be in touch to determine which workgroup you will be part of.

**Dr. Tierney** introduced guest speaker and council member **Julie Gortze**, Julie is a registered nurse who spent most of her career working in pediatric home care, subacute care and acute care. Julie has personal experience with complex medical conditions and has learned firsthand what patients and families deal with while searching for a cause and receiving a diagnosis that has no cure. Julie is the founder and president of Rare New England. She will be presenting information about the rare disease NE data mapping project.

The purpose of the data mapping project is to begin to generate the geographic data needed to draw a map of rare disease in NE. The primary objective is to collect location data from as many rare disease patients and families as possible and then map it by zip code to create a snapshot of where rare disease impacts NE.

Julie told the council that in 2021 Rare NE received a grant. In 2022 they held focus groups to determine how best to move forward with the grant. In 2023 Rare NE decided to create a map of prevalence of rare disease in NE. They focused on this because, they want to be able to inform legislation and the medical community with a good estimate of how many individuals are living with this specific diseases.

The survey, launched last week. We are keeping it simple. Asking for zip code, diagnosis, mutation (What disease do you have and where do you live). A benefit of the focus groups was the connection of patients and their families to each other, a network of support groups. A challenge of prevalence is that the data is not accessible. The concept of identifying clusters would be part of the data collected.

**Dr. Tierney** spoke about a potential collaboration of the RDAC (Work Group #1) collaborating with Rare NE. He asked how duplication of data could be managed. Rare NE is not that far into the data collection to know about the researchers and statisticians involved in the project.

The group talked about when people move, which often happens with those with rare diseases. They may move to be near their rare disease specialist was discussed.

**Dr. Tierney introduced Guest Speaker Yue Huang,** a senior research analyst at the Massachusetts Health Policy Commission, an independent state agency charged with monitoring healthcare spending growth and providing data-driven policy recommendations regarding care delivery and payment reform. Ms. Huang’s work utilizes large, quantitative datasets to study topics such as prescription drug spending, drug coupon use, surprise billing, and emergent modalities of care including urgent care centers and telehealth. She has an MS in food policy and applied nutrition from the Friedman School of Nutrition at Tufts University.

Yue will be presenting the Health Policy Commission analysis on the increased role of telehealth services in Massachusetts.

The Massachusetts Health Policy Commission (HPC), was established in 2012, as an independent state agency charged with monitoring healthcare spending growth in Massachusetts and providing data-driven policy recommendations regarding healthcare delivery and payment system reform. The HPC’s mission is to advance a more transparent, accountable, and innovative healthcare system through independent policy leadership and innovative investment programs.

The discussion focused on pay parity. Telehealth for mental health remains high but for other conditions, the medical community has largely returned to seeing their patients in person. Dr. Tierney presented the concept of studying the relationship between telehealth and health outcomes.

**Celia Segel** raised the issue of telehealth improving access for patients who need to frequently see many specialists to manage complex conditions, and need to travel long distances to see multiple different providers.

**Dr, Tierney** asked if there were any announcement. No one shared any announcements.

Dr. Tierney shared that the last member will be appointed soon.

**NEXT FULL COUNCIL MEETING: January 19, 2023 9:00 am – 11:00 am**

Tai Pasquini made a motion to adjourn

Glenda Thomas seconded the motion.

Dr. Tierney asked if all were in favor of adjourning.

**Meeting adjourned at 10:56**

**WORKGROUPS Volunteers via chat**

**Workgroup 1**

D. Miller

A. Mahady

A. Dwyer

A. Lane

T. Pasquini

A. Wessel

**Workgroup 2**

Jay Livingstone

J. McNary

L. Deck

**Workgroup 3**

L. Joseph

G. Thomas

S. Patel

J. McNary