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

**MEETING MINUTES**

REMOTE MEETING:    Thursday, May 18, 2023, 9:00-11:00 AM

*Approved July 20, 2023*

**Welcome – Dr. Dylan Tierney** welcomed all to the meeting.

He proceeded with a guest participant announcement letting all know that 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. He asked all guests on the call to please ensure that their video is turned off and their audio is on mute.

**D Tierney conducted a roll call to establish a quorum.**

|  |  |  |
| --- | --- | --- |
|  | **Council Member** | **Present** |
| 1 | Charlotte M. Boney, M.D | Joined at 10 |
| 2 | Janis Creedon | - |
| 3 | Lisa Deck | X |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | X |
| 5 | Senator Paul R Feeney (Maya Rabinovitz) | X |
| 7 | Julie D. Gortze, RN | X |
| 8 | Guadalupe Hayes-Mota, MBA, MS, MPA | X |
| 9 | Lena Joseph, RN, CPN | X |
| 10 | Representative Hannah Kane (Sophia Flionis) | X |
| 11 | Andrew A. Lane, MD, PhD | X |
| 12 | Representative Jay Livingstone (Daniel Butler) | X |
| 13 | Jeff R. Livingstone, PhD | - |
| 14 | Diane Lucente, MS, LCGC | X |
| 15 | Alexsandra B. Mahady | Joined at 9:30 |
| 16 | Jenn McNary | X |
| 17 | David T. Miller, MD, PhD | - |
| 18 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | X |
| 19 | Shivang Patel, Pharm.D. | X |
| 20 | Asma Rashid, MS, CGC | X |
| 21 | Michele Rhee, MBA, MPH | X |
| 22 | Robert E. Schultz, MBA | X |
| 23 | Celia Segel, MPP | Joined at 9:40 |
| 25 | Glenda E. Thomas | X |
| 26 | Ryan Thompson, MD, MPH | X |
| 27 | Dylan Tierney, MD, MPH | X |
| 28 | Ann Wessel, MS, RD, LDN | X |
| 29 | Ross Zafonte, DO | - |

A quorumwas established, **D Tierney** brought the meeting to order at 9:05

**D Tierney proceeded to the first order of business, Voting** to approve the meeting minutes from the last full council meeting on 3/16/23. He asked if all had received the minutes and then asked if there were any edits or corrections to the minutes.

**A Dwyer** stated that there was a correction on Pg 4 of the minutes; instead of “Some states had good data,” it should read “Some Nordic countries have good data.”

**D Tierney** asked if there were any other corrections. Can I have a motion to vote on the minutes with the correction identified by A Dwyer?

**A Dwyer** motioned to vote on the minutes with the corrections as noted.

**T Pasquini** seconded the motion.

**D Tierney** then conducted a roll call vote to approve the minutes from 3/16/23

|  |  |  |
| --- | --- | --- |
|  | **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 | Senator Paul R Feeney | X |
| 7 | Julie D. Gortze, RN | X |
| 8 | Guadalupe Hayes-Mota, MBA, MS, MPA | X |
| 9 | Lena Joseph, RN, CPN | X |
| 10 | Representative Hannah Kane (Sophia Flionis) | X |
| 11 | Andrew A. Lane, MD, PhD | X |
| 12 | Representative Jay Livingstone | X |
| 13 | Jeff R. Livingstone, PhD | - |
| 14 | Diane Lucente, MS, LCGC | X |
| 15 | Alexsandra B. Mahady | - |
| 16 | Jenn McNary | X |
| 17 | David T. Miller, MD, PhD | - |
| 18 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | X |
| 19 | Shivang Patel, Pharm.D. | X |
| 20 | Asma Rashid, MS, CGC | X |
| 21 | Michele Rhee, MBA, MPH | X |
| 22 | Robert E. Schultz, MBA | X |
| 23 | Celia Segel, MPP | - |
| 25 | Glenda E. Thomas | X |
| 26 | Ryan Thompson, MD, MPH | X |
| 27 | Dylan Tierney, MD, MPH | X |
| 28 | Ann Wessel, MS, RD, LDN | X |
| 29 | Ross Zafonte, DO | - |

**D Tierney introduced the Guest Speaker**: **Representative Dylan Fernandes**

Representative Dylan Fernandes has served Falmouth, Martha’s Vineyard, and Nantucket in the legislature since 2017. He has focused on combating climate change, advancing affordable housing, and addressing the opioid and heroin epidemic. He serves as the Vice Chair of the Committee on Environment and Natural Resources. He has seats on the Committee on Ways and Means, the Committee on Telecommunications, Utilities & Energy, and the Committee on Global Warming and Climate Change.

A strong believer in addressing inequality, Rep. Fernandes got his start in politics as an organizer for Elizabeth Warren in 2012. He served as Maura Healey’s political director on her campaign for Attorney General. He worked in the Attorney General’s Office fighting discrimination in the Civil Rights Division and later served as digital director for the office.

A fourth-generation Cape & Islands resident and the son of two small business owners, his great-grandparents immigrated to the United States from the Azores before settling in Falmouth to work as strawberry farmers. Fernandes received his bachelor's degree with a double major in Economics and Political Science from the College of Charleston and a Master’s of Public Administration from Harvard University, where he was a Rappaport Scholar.

**Rep Fernandes** thanked the group for having him. He stated that he had been diagnosed with a rare disease in his teens. His journey began in adolescence. He would develop severe pain after physical activity. He was into sports so pain started to interfere with his ability to play sports. At 14 he went through a series of tests and a large mass was found in his leg. He was diagnosed with a vascular anomaly and he received his first surgery at age 15. After surgery he got some relief but within a short time, the pain was back. A few years later, the surgery was done again, however the results were the same. He only had short-lived relief from the pain. In 2014, he went to see Dr. Alomari at Boston Children’s Hospital. Dr. Alomari was known for his treatment of vascular anomalies. Dr. Alomari explained that he had just discovered a rare disease that could explain his pain. Over the next several years, his pain became debilitating, often affecting his ability to walk. Around this same time, he stated that he decided to run for office as state representative. Days on the campaign trail were very long and tiring. He stated that he would often have to come home and lay down. It had started to take a serious mental toll on him. He then started looking up the diagnosis on the internet and found some Facebook groups of people with the same diagnosis. It was really difficult to read some of this information. He states that he became a bit hopeless reading the stories of others with the same diagnosis. A short time after that, Dr. Alomari called to tell him that removing the growth could improve his pain. He then underwent surgery to remove the lesion from his leg. The doctor told him that his prognosis was uncertain and that the growth could come back. Rep Fernandes stated that the surgery had been a success, and he has been pain-free ever since. Within 9 months after the surgery, he was back running and was able to run a 5K. This was a huge milestone for his mental health. He hopes that he will remain disease free, but he really doesn’t know. He states that he feels very fortunate that his disease was in a place that could be removed. Some of the people that he has talked to or read about are not so lucky. Their disease is in places where surgery is not possible. As an elected official, he received a lot of press going through this. He now realizes that as an elected official, he can help in ways that others cannot. He is willing to support the work of the council in any way he can. He stated that he would always be a champion for the work of the rare disease advisory council.

**D Tierney** thanked Rep Fernandes for talking about his rare disease experience. He stated that it was nice to give people hope. He then asked how this experience might impact his work as a legislator.

**Rep Fernandes** stated that this journey has been very difficult and emotional. I know I am more empathetic when people tell me their stories. I’m mindful that many people are going through something. I never really talked about what I was going through when I was in such severe pain and others may be doing the same. As a legislator, this is a very difficult time in healthcare in Massachusetts right now. We want to fund the large teaching hospitals because they are often the places where the innovation is happening on the other hand, it is very expensive for them. We also know we need to support the small community hospitals. He reminded all that he is from the cape, where small hospitals are a vital part of the community.

**D Tierney** thanked him and asked if anyone else had any questions.

**J McNary** thanked Rep Fernandes and stated that she was the mom of 3 children with a rare disease. She stated that she had 2 adult sons with Duchenne Muscular Dystrophy. She then asked what Rep Fernandes thought about the value of early diagnosis.

**Rep Fernandes** stated that he didn’t know how to respond to that. Although he was misdiagnosed, his disease wasn’t even discovered yet.

**L Joseph and J Gortze** both thanked Rep Fernandes for speaking about this journey.

**T Pasquini** asked Rep Fernandes how he thought that the group could capture the burden of disease? She stated that she knows that he states that he is doing great now, but she was curious about the disease's impact on his life.

**Rep Fernandes** stated that he thought that his mental health was affected the most. The disease journey really takes a toll on your ability to be positive and have hope. He stated that although he is still doing great now, there are times when the mental impact affects his day to day.

**J McNary** then asked Rep Fernandes what he thought was the most important issue for the residents of Massachusetts with a rare disease.

**Rep Fernandes** stated that he felt that there was a strong need for kids to network with others going through the same thing they are. In fact, there is probably a need for parents to network with other parents also.

**D Tierney** thanked Rep Fernandes for his time and his vulnerability to share his experience.

**D Tierney** then said we would move on to the next agenda item, WorkgroupUpdates. He asked if Workgroup 1 wanted to start, and then he expected that Workgroups 2 and 3 would be reported together.

**Workgroup 1 – Tai Pasquini** noted that D Miller, the workgroup chair, was traveling to a graduation celebration so she would report for the group. As you know, the workgroup has been working on writing a white paper to support our estimate for prevalence of rare disease in Massachusetts. We are not ready to share our paper but we have finalized our outline. (she then showed the outline) She stated that the literature discusses that approximately 3-7% of the population have a rare disease however, this does not include many diseases, like rare cancers and other known rare diseases. Therefore, our group has discussed ranges. We think that estimating a range may be more appropriate. She then walked through the outline and explained the thought process for estimating the prevalence number in Massachusetts.

**D Tierney** asked how the group will decide on the comparisons to use.

**T Pasquini** stated that the group wanted to get to the most accurate number by comparing population estimates for that specific disease. Some diseases have really good data, so we can compare those data sets to Massachusetts population data. We think we can look at the most common diseases that have good data and verify if Massachusetts data is reflective of the numbers they report. Using this method, we can estimate the number of rare diseases in Massachusetts with some confidence.

**D Tierney** asked if the group would use hard data to compare to a mathematical conclusion.

**A Dwyer** stated that the conditions and diseases the group selected to review, were the ones with the best data.

**D Tierney** asked if the group didn’t have hard numbers on a specific disease, how would they do come up with an estimate for that disease?

**T Pasquini** stated that they would be using the Massachusetts cancer registry and the Massachusetts newborn screening data for comparisons, but all diseases are not accounted for so the group would have to make some estimates to get to the statewide number for all rare diseases.

**A Lane** stated that the group was working really hard to get to a number that can be verified. The group has spent a lot of time discussing this issue. Although this sounds like an easy question to answer, it isn’t. We are working hard to ensure that we can justify the number we come up with. We are also thinking about using ranges vs a specific number.

**T Pasquini** reiterated that the task at hand is very complex. We now know why other states have not done this. We are working hard to have a white paper draft before our next full council meeting in July.

**D Tierney** then asked if Workgroups 2 and 3 wanted to report on their group's progress.

**J McNary** stated that she would start. She let everyone know that the Workgroup 2 (W2) and Workgroup 3 (W3) decided to work together. They were both working on surveys to gather data and realized that it may be helpful to work together. Michele Rhee and Shaving Patel have been working together to develop a survey encompassing all the questions from W2 and W3. Jenn asked if either Michele or Shaving would like to give an update on their progress.

**M Rhee** stated that she would start but it was really Shaving that has been putting in the work to create a survey that could work. She stated that they created a tool allowing a participant to open the survey and decide what information they wanted to input. We are currently working through the structure of the survey with the workgroups.

**S Patel** added that he was working on the details to ensure a smooth transition between sections. He hoped to have a draft of the survey by next week. The groups would then review the survey. After the review, the workgroups would beta-test the survey with the members. Once this is done, we will send it to the full council.

**D Tierney** thanked the workgroups for their hard work. He acknowledged that everyone has a full-time job and all this work is over and above everyone’s other obligations. He thanks all for their dedication to this work.

**J McNary** asked if the workgroups realized that they would be coming up with recommendations to send to the governor and legislature but should they also think about what else may be needed to carry out the recommendation, such as a person or budget.

**D Tierney** noted that we all realize that there may be more questions coming out of our work, but that was a good thing.

He then proceeded to move on to the next agenda item. He introduced the next speaker, Dr. Chuck Pu from MassHealth Long Term Services and Supports Division.

**Dr. Chuck Pu** was introduced as the senior medical director of the MassHealth Long Term Services and Supports. He spoke to the council about MassHealth's work to support those with a chronic condition that requires extra support. He started by talking about the healthcare spending in the US, noting that the US spends more on healthcare than any other developed country in the world and in some cases we are the lowest performing. He then shifted to medical spending and social care spending. Social care spending is often not included in the whole picture of health care spending. He stated that if we looked at both the medical healthcare spending and added social care spending together, the US would be more in the middle. He summarized by saying that the US is often reactive site-specific, and time driven vs other countries that look at the whole person and the needs of the whole person, not just what is needed for a specific disease or condition.

Dr. Pu then shifted to talk about MassHealth Long Term Services and Supports (MHLTSS). What does MHLTSS do? He provided a summary document (listed in meeting support materials) describing the services MHLTSS provided. He reviewed his slides and ended by asking if there were any questions.

**J McNary** stated that as a mother of 3 children with a rare disease and their advocate, she has been struck by the gaps in services that are provided to adults with a rare disease vs elderly residents. She stated that her adult children with a rare disease, often struggle to find and receive services, even when they qualify for them. There are clear gaps in services that are needed.

**C Pu** thanked J McNary for sharing her story and stated that he appreciated her feedback. He said he would be happy to work with the group to identify the existing gaps in services. He added that he knows that there is a vital need for care coordination. If there is an opportunity to improve, he would like to work on that.

**T Pasquini** thanked C Pu for his talk and stated that some feedback from her workgroup was related to the need for experienced case managers. She received feedback from patients stating that the case managers don’t understand their rare disease.

**C PU** stated that he understood that the case managers in MHLTSS often focus on the patient's functional needs and not necessarily on the disease itself. He stated that he would like to learn more about this. He stated that most of the case managers in MHLTSS come from the social services side and not necessarily the medical side. We need to understand this issue better.

He then stated that there was a huge workforce shortage issue. We are trying our best to support the programs we currently have first. When there has been an increase in funding, we have used that money to strengthen our existing programs. For example, we just provided rate enhancements by increasing reimbursement rates. This was long overdue. We know there are areas that need additional resources. Our home enhancement program is an example.

**D Tierney** asked how people with rare diseases could have their voices heard.

**C PU** stated that advocacy groups and other stakeholders play a large role in funding priorities.

**C Segel** asked if there had been any discussions around private payers. How do people with private insurance access these services.

**C Pu** stated that all of these services are only available for MassHealth clients. Many social supports for people on private insurances are paid out of pocket. Until we move toward outcome based healthcare, unfortunately, many private payers don’t pay for the wrap around and social services that people need to be live a quality life. Often times, people on private insurance have to spend down all their money to pay for these services, then they qualify for MassHealth and can become eligible for our services. I so wish we all had wrap around care but we don’t.

**D Tierney** thanked C Pu for his talk and important information. He then said he would be moving on to the next agenda item. Announcements. He stated that he had a few announcements then he would open it up to the group. D Tierney announced that the vacant positions of Dr. Michael Green and Dr. Michael Sherman were in the process of being filled.

He then stated that we were working on a plan to hold a public town-hall-style meeting to gather public input about our work. He stated that it was decided to hold this meeting virtually. This would ensure that the greatest number of people are able to participate. We are working with W2 and W3 to help structure this meeting. We are planning for this meeting to occur in the fall.

He then added that he was invited to attend and participate in a panel discussion at the NORD Breakthrough Summit on Oct 15-17 in Washington, DC. He said the NORD is offering funding to Massachusetts RDAC to support 3 members to attend. HE asked if anyone would like to attend; they should contact him and let him know they want to go.

He asked if there were any other announcements.

He then asked if there was a motion to adjourn.

**Workgroup 2 – Chair Jenn McNary**

***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

**DISCUSSION**

**Workgroup 3 – Chair Lena Joseph**

***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.

**DISCUSSION**

**MassHealth Long-Term Services and Supports Presentation**

MassHealth Office of Long-Term Services and Supports (OLTSS) 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.

**MassHealth LTSS speaker – Chuck Pu**

Charles (Chuck) T Pu, MD, FACP, CMD

Senior Medical Director **|** EOHHS MassHealth Office of Long-Term Services & Supports

For Health Consulting **|** UMass Chan Medical School

Attending Physician, Mass General Hospital Division of Palliative Care & Geriatric Medicine

Assistant Professor, Harvard Medical School.

**ANNOUNCEMENTS**

**NEXT FULL COUNCIL MEETING: July 20th, 2023, 9:00 am – 11:00 am**

**M Rhee** motioned to adjourn. **T Pasquini** seconded.

**D Tierney** adjourned the meeting at 10:57.