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

**Meeting Minutes**

Approved March 28, 2024

REMOTE MEETING:    Thursday, January 18, 2024, 9:00-11:00 AM

**Dr. Dylan Tierney** welcomed everyone to the meeting.Then conducted

Roll Call to establish a quorum.

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|  | **Council Member** | **Present** |
| 1 | Charlotte M. Boney, M.D | X |
| 2 | Janis Creedon | - |
| 3 | ~~Lisa Deck~~ (resigned 11/23) | - |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | X |
| 5 | Senator Paul R Feeney (Maya Rabinovitz) | X |
| 6 | Julie D. Gortze, RN | - |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA | X |
| 8 | Lena Joseph, RN, CPN | X |
| 9 | Representative Hannah Kane (Kerry Shea) | X |
| 10 | Andrew A. Lane, MD, PhD | X |
| 11 | Representative Jay Livingstone | X |
| 12 | Jeff R. Livingstone, PhD | X |
| 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) | - |
| 18 | Shivang Patel, Pharm.D. | X |
| 19 | Asma Rashid, MS, CGC | X |
| 20 | Michele Rhee, MBA, MPH | X |
| 21 | Robert E. Schultz, MBA | X |
| 22 | Yue Huang, MS | X |
| 23 | Gail Ryan, PharmD | X |
| 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 brought to order at 9:05 am.

**D Tierney** asked all if they had received the minutes from the last full council meeting. All agreed. He asked if there were any edits or corrections. No one made any suggestions.

**D Miller** made a motion to accept the minutes as presented.

**G Thomas** seconded the motion.

**D Tierney conducted a roll call vote to accept the minutes from council meeting on November 16th as presented.**

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|  | **Council Member** | **Accept** |
| 1 | Charlotte M. Boney, M.D | X |
| 2 | Janis Creedon | - |
| 3 | ~~Lisa Deck~~ (resigned 11/23) | - |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | abstained |
| 5 | Senator Paul R Feeney (Maya Rabinovitz) | X |
| 6 | Julie D. Gortze, RN | - |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA | X |
| 8 | Lena Joseph, RN, CPN | - |
| 9 | Representative Hannah Kane (Kerry Shea) | X |
| 10 | Andrew A. Lane, MD, PhD | X |
| 11 | Representative Jay Livingstone | X |
| 12 | Jeff R. Livingstone, PhD | abstained |
| 13 | Diane Lucente, MS, LCGC | X |
| 14 | Alexsandra B. Mahady | X |
| 15 | Jenn McNary | abstained |
| 16 | David T. Miller, MD, PhD | X |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | - |
| 18 | Shivang Patel, Pharm.D. | X |
| 19 | Asma Rashid, MS, CGC | X |
| 20 | Michele Rhee, MBA, MPH | abstained |
| 21 | Robert E. Schultz, MBA | abstained |
| 22 | Yue Huang, MS | X |
| 23 | Gail Ryan, PharmD | X |
| 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 | - |

**D Tierney** then introduced our Guest Speaker, Rachel Bennett.

Rachel is an advocate and a parent of a child with a rare disease. She is the Director of [CVI Now](https://www.perkins.org/cvi-now/), a Parent Advocacy and Support network at the CVI Center at Perkins School for the Blind.  She has a son, Henry, with a rare disease, an extremely rare variant of the ASTN-1 gene.  She and her family relocated from out of state to MA in order to get care for Henry.  Henry also has CVI, which is common within the rare disease community, and it frequently goes undiagnosed.  Rachel can share Henry's story, as well as explain how frequently this is undiagnosed.

Rachel first thanked the council for giving her an opportunity to speak. She let us know that CVI is the leading cause of blindness in kids. She followed by saying that she first wanted to talk about Henry, her son. He is eleven years old and is in the 5th grade. She stated that one of the hardest things as a parent with a rare disease is always trying to prove why he needs help. To tell us a little about our journey, she said that she had a normal delivery, but at about three months, she noticed his eye quivering. They started seeing a lot of specialists to figure out what was wrong. Henry was also failure to thrive and had a hard time gaining weight.

At age 2, Henry received genetic testing, and she knows they were fortunate to have insurance pay for this testing when often they do not.

At 3 or 4, the ophthalmologist diagnosed him with a behavioral disorder. In hindsight, that was very frustrating. It wasn’t until his preschool teacher identified that he was not using his eyes. After that, we saw more doctors, and he was finally diagnosed with CVI at age 5. Once we had this diagnosis, we looked for a place where we could find experts in CVI. This was when we decided to move to Boston. We know how critical it is to diagnose CVI early. Henry’s access to educational services, social supports, and resources is drastically different than in Maryland, where we lived when he was born. We feel strongly that children should be screened for CVI between birth and age 1. So, many children are misdiagnosed or not diagnosed at all. She then stated that she would stop but would be happy to answer any questions.

**D Tierney** thanked Rachel for sharing her story with the council and asked if anyone had a question.

**J McNary** thanked her for sharing and asked if she was working on any legislative actions that the council should be aware of and how she is managing care for Henry.

**R Bennett** responded by saying that she felt that CVI screening should be routine for kids. She stated that NCI is starting to do research on CVI and has created a data registry. One challenge is that there is no definitive ICD code for CVI. She is following their work and will let the council know if there is any information that is pertinent for the council to know. She then added that care is challenging. This question is hard to answer. One of her biggest challenges is managing all the doctor appointments for Henry. It is even more challenging for us, and I’m sure many people with a rare disease because you don’t know what is next. It’s hard to plan for a symptom or outcome when you have no idea it’s coming or expected.

**G Ryan** asked if she has been able to communicate with others with CVI diagnosis.

**R Bennett** expressed that she was very lucky that she worked in a job that she loves, and it allows her to communicate with other families in a similar situation. It’s also important to know that research shows that there are substantially more children with CVI than we know. It is underdiagnosed.

**A Dwyer** Thanked her for her advocacy and passion for helping others with CVI

**D Tierney** Thanked her for taking the time to talk with the council. He then asked if all had received the annual report.

All agreed. He then put up the executive summary on the screen and asked if anyone had any edits or thoughts on the paragraph.

**J McNary** suggested adding information about the speakers we have at our full council. Some of the information they have shared is important to put in this report.

**D Tierney** suggested that Mary Lou could work with Jenn to get this information into the report.

**G Ryan** added that she wondered if we were underestimating the number of Massachusetts residents with a rare disease. She added the NORD estimated that the number was more like 10% of the population. They estimated that between 3.5 and 6% of the MA population was affected. **D Tierney** stated that the prevalence workgroup extrapolated data from several registries as opposed to just using the NORD estimate. They wanted to be able to substantiate the number they presented.

**D Tierney** stated that we never expected to be able to come up with a specific number. He asked if anyone from the prevalence group wished to speak.

**D Miller** asked if the group felt that the number estimate was too precise?

**A Lane** stated that he was on the workgroup and that he recommended that we use the rounded number on both sides of the spectrum.

**D Tierney** stated that he would rework the paragraph on prevalence and asked if workgroup 1 could review it prior to finalizing the report.

**D Miller and A Lane** both agreed to review Dylan’s rework and also share with other workgroup members.

**D Tierney** asked all to send any edits or revision recommendations to him by Friday, January 26th. He stated that Mary Lou would send out a reminder on Thursday.

**Jeff Livingstone** asked if the public could have access to the report and the testimonies of the public.

**D Tierney** stated that both reports would be available to the public on our website. He then asked Rep. Livingstone if he thought that the report could wait until our next meeting to finalize. That would mean our report would not be submitted to the legislature until March.

**Rep Livingstone** responded by saying that we should submit our report timely, but it would be accepted if we submitted it late. Another option is to revise the legislative language so that there is enough time to summarize the year’s work prior to submission.

**D Tierney** asked to move on to the next agenda item. He discussed the last steering committee meeting. He stated that Guadalupe Hayes-Mota joined the group in December. The committee discussed priorities for the council in 2024, and two themes emerged.

* Collecting data that will strengthen the council’s understanding of the rare community
* Establishing a case for funding an office of rare diseases in state government

He added that we had a productive year by creating a survey and conducting a public town hall, but we need to do more in 2024 in order to gain a better understanding of who is part of the rare disease community and what their needs are.

He also stated that the steering committee discussed other issues like home care and telehealth but felt that we should focus on the broader issues versus focusing on a specific topic.

**J McNary** asked if we could discuss working on a budget and fundraising. We know that there is a lot of work to gather this data, and funding would help us hire someone to help.

**G Thomas** asked if we could spend time looking at legislative bills and how we may be able to advocate for those that are important to the rare community.

**D Tierney** stated that it may be helpful to have a workgroup that focuses on legislation.

**J McNary** asked if the RDAC could take a position on legislation. If we can’t take a position, can we at least make a statement about why the legislation is important to the rare community?

**D Tierney** asked G Thomas if she was willing to take the lead on this?

**J McNary** added that she would be willing to work with Glenda on this.

**G Thomas** stated that she welcomed J McNary’s help and would take the lead.

**D Tierney** stated that he would summarize the discussion and stated that the council would focus on three priorities for 2024.

1. Expanding the foundational work of gathering data about the rare disease community in Massachusetts
2. Increase awareness about the Rare Disease Advisory Council and its work
3. Tracking and monitoring legislation relevant to the rare disease community

D Tierney then asked for an update from workgroups 2 and 3.

**J McNary stated** that she would speak for both groups as L Joseph was in clinic today. She stated that booth chairs agreed to dissolve the workgroups as they currently exist. The workgroups accomplished their goal of creating the survey, but now we needed to figure out how to market it to get more responses. The responses to date are very small. She would like to call this new workgroup the Community Engagement Planning workgroup. She asked for any volunteers who are willing and able to put in some time on the projects at hand. The focus for this workgroup would be to:

* Increase awareness of the RDAC
* Collect data about the rare disease community (through surveys, testimonies and small group interviews)
* Develop a report for the legislature

She asked if anyone had any connections with industry or business partners who may be able to help develop a marketing plan for the RDAC. If so, could they please reach out to her? She then asked if anyone would like to work on this to please reach out to her.

**Y Huang** asked if the Office of Rare Disease was expected to replace the RDAC.

**D Tierney** stated that the steering committee thought that the office would be staffed and would be charged with implementing the recommendations of the RDAC. The RDAC would remain as an advisory body.

**Y Huang** thanked DT for the clarification and stated that she thought this made a lot of sense.

**D Tierney** then added that most members' terms would be expiring in 2024. He stated that Mary Lou would be reaching out to all once we had an understanding of the renewal process. He asked all to think about continuing on the council but realized that sometimes life gets in the way. He asked that any member not renewing to consider staying on the council until there is a replacement.

He then announced that there would be a legislative briefing on Tuesday, February 13th. It was not a public meeting, but all RDAC members were welcome to attend. If anyone plans on attending, they should reach out to Mary Lou to let us know who will be present. The purpose of this briefing is to inform as many legislators as possible about the RDAC and its work to date. Rep. Kane sent out a save the date to legislators on behalf of all legislative representatives and senators on the council.

**G Thomas** asked if we could take a few minutes to discuss our website. She expressed that we do not have any way for the public to reach out to us.

**D Tierney** thanked G Thomas for her concerns and stated that we have discussed in the past but have not come up with a plan on how to do this responsibly. He asked if Jen’s new workgroup could address this?

**J McNary** stated that she planned on discussing this very issue in her workgroup and welcomed G Thomas to join the workgroup.

**D Tierney** then asked if anyone had any announcements. No one added so he asked if there was motion to adjourn.

**G Ryan** made a motion to adjourn. **A Dwyer** seconded the motion.

**D Tierney** adjourned the meeting at 10:30 am.

**9:30 VOTE to accept minutes from last full council meeting on November 16, 2023**

**9:45 Annual Report review and discussion. All received the final draft. I have not had any recommendations for revisions or edits.**

**VOTE to approve 2023 Annual Report and submit to legislature**

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|  | **Council Member** | **approve** |
| 1 | Charlotte M. Boney, M.D |  |
| 2 | Janis Creedon |  |
| 3 | ~~Lisa Deck~~ (resigned 11/23) |  |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN |  |
| 5 | Senator Paul R Feeney |  |
| 6 | Julie D. Gortze, RN |  |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA |  |
| 8 | Lena Joseph, RN, CPN |  |
| 9 | Representative Hannah Kane |  |
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| 12 | Jeff R. Livingstone, PhD |  |
| 13 | Diane Lucente, MS, LCGC |  |
| 14 | Alexsandra B. Mahady |  |
| 15 | Jenn McNary |  |
| 16 | David T. Miller, MD, PhD |  |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) |  |
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| 28 | Ross Zafonte, DO |  |

**10:00 – 10:30 Setting priorities for 2024**

**The steering committee met last month and discussed priorities for 2024. They felt strongly that we should focus on building the case for establishing an office of rare disease. Defining the prevalence was the first step but we have a lot more information and data to collect before we will have a clear understanding of the needs of the rare disease community. The committee felt that the first priority was to develop a marketing plan. Workgroups 2 & 3 will be discussing that next. The topics discussed included:**

* Developing a marketing plan
* Needs assessment
  + Other public forums
  + Qualitative interviews with key stakeholders
* Legislative tracking
* Legislative briefing
* Strategic Planning
  + Office of Rare Disease
  + Ombudsman
  + Helpline

The steering committee thought that forming a few new workgroups could be helpful, but we needed to recognize that most members did not have the time to do the time-consuming data collection and information gathering that was needed to support our cause.

They thought we should focus on 2-3 topics for the year.

Ask for a discussion about the council's direction in 2024. We also talked about investigating home care issues but didn’t confirm that it should be one of the topics to focus on in 2024.

**10:30 – 10:55 Workgroup 2 & 3 update by Jenn and Lena**

Jen and Lena will give an update on their workgroup’s progress.

**Review Council Membership renewal– Most members will be coming to the end of their terms in 2024. Mary Lou will be reaching out to each of you, if she hasn’t already to ask if you would like to remain as a member of the council. If you wish to remain on the council there will be a process for your renewal and Mary Lou will let you know what is needed. If you cannot or would not like to continue as a member of the council, we realize that life changes and appreciate all of your work to date. The process sometimes takes time so we ask that you remain on the council until your replacement is named.**

**Meeting Schedule for 2024**

**The meeting schedule will remain the same in 2024. The full council will meet every other month on the third Thursday of the month from 9:00 – 11:00 am beginning in January.**

**The Steering Committee will meet every other month on the third Thursday of the month from 10:30 – 11:30 am beginning in February.**

**ANNOUNCEMENTS: Legislative Briefing will be held on**

**TUESDAY, FEBRUARY 13th at 1:00 pm in Senate Room 428**

**This is our opportunity to share our work with the legislature. Rep. Kane sent out an invite from all of the legislators on the council. All council members are welcome to attend. Please let Mary Lou know if you plan to attend. This is not a public event.**

**NEXT FULL COUNCIL MEETING: is scheduled for March 21st, 2024, 9:00 am – 11:00 am. Due to a conflict, we need to reschedule to either the week before (March 14th) or the week after (March 28th)**

**Ask for all who can make the 14th to raise their hand. ( I will tally)**

**Then ask for all who can make the 28th to raise their hand.**

**Once confirmed announce the new date.**

**ADJOURN**

Ask for a motion to adjourn.

Repeat the name of the person who makes the motion and the person who makes the second.

Ask all if they agree to adjourn.

**Adjourn** – State time meeting is adjourned for the minutes.