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

**Meeting Minutes**

**Approved May 18, 2023**

REMOTE MEETING:    Thursday, March 16, 2023, 9:00-11:00 AM

**Welcome: D Tierney** welcomed all to the meeting. He then announced that the public was always welcome to join any of the RDAC meetings; however, only guests that were on the agenda would be allowed to speak during the meeting. He asked all guests on this call to please make sure that their video is turned off and their audio was on mute.

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

**D Tierney** let everyone know that there was a slight change to the agenda. Representative Dylan Fernandes was unable to attend.

He also wanted to inform everyone that the council’s final member appointment was made. Senator Paul Feeney was newly appointed to the council. He asked if Senator Feeney would introduce himself.

**Senator Feeney** introduced himself and said he was excited to be a council member. He looked forward to working with everyone and participating in the great work of the council.

**D. Tierney t**hankedSenator Feeney and welcomed him to the council. He then stated that he wanted to inform everyone that council member Celia Segel would be on leave for a few months, but another Health Policy Commission member would fill in for her in her absence. He asked Katherine Mills to introduce herself.

**K Mills** introduced herself as Senior Director for Marketing oversight and transparency at the Health Policy Commission. She said she was excited to fill in for C. Segel in her absence.

**D Tierney** thanked K. Mills and welcomed her to the council and stated that he would conduct a **Roll Call to establish a quorum**

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|  | **Council Member** | **Present** |
| 1 | Charlotte M. Boney, M.D | no |
| 2 | Janis Creedon | no |
| 3 | Lisa Deck | no |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | yes |
| 5 | Senator Paul R Feeney | yes |
| 6 | Michael R. Green, M.D., Ph.D. | no |
| 7 | Julie D. Gortze, RN | yes |
| 8 | Guadalupe Hayes-Mota, MBA, MS, MPA | yes |
| 9 | Lena Joseph, RN, CPN | yes |
| 10 | Representative Hannah Kane | yes |
| 11 | Andrew A. Lane, MD, PhD | yes |
| 12 | Representative Jay Livingstone | yes |
| 13 | Jeff R. Livingstone, PhD | yes |
| 14 | Diane Lucente, MS, LCGC | yes |
| 15 | Alexsandra B. Mahady | yes |
| 16 | Jenn McNary | yes |
| 17 | David T. Miller, MD, PhD | no |
| 18 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | yes |
| 19 | Shivang Patel, Pharm.D. | yes |
| 20 | Asma Rashid, MS, CGC | no |

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| 21 | Michele Rhee, MBA, MPH | no |
| 22 | Robert E. Schultz, MBA | no |
| 23 | Celia Segel, MPP (Kate Mills) | yes |
| 24 | Michael Sherman, MD | yes |
| 25 | Glenda E. Thomas | no |
| 26 | Ryan Thompson, MD, MPH | yes |
| 27 | Dylan Tierney, MD, MPH | yes |
| 28 | Ann Wessel, MS, RD, LDN | yes |
| 29 | Ross Zafonte, DO | yes |

**Quorum was established. D Tierney brought the meeting to order at 9:09**

**A Lane** notified all that Council Member Dr. Michael Green passed away in February. He shared the obituary (below) The sad news silenced all.

<https://www.telegram.com/obituaries/pneo0429433>

**D Tierney** thanked A Lane for sharing with the group and stated that he will be missed.

**A Lane** shared that he knew Dr. Green before working with him on the council and stated that he was a fierce advocate for rare diseases and hoped that the council could be empowered to carry on his work.

**D Tierney** asked if everyone received the minutes from the last full council meeting on January 19th. All stated yes. He then asked if anyone had any edits or corrections. No one responded. He then asked if there was a motion to accept the meeting minutes from January 19th as presented.

**T. Pasquini** made a motion to accept the minutes

**A Lane** made a second to the motion.

**D Tierney** stated that he would conduct a role call to accept the minutes from January 19th.

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|  | **Council Member** | **approve** |
| 1 | Charlotte M. Boney, M.D | Not present |
| 2 | Janis Creedon | Not present |
| 3 | Lisa Deck | Not present |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | yes |
| 5 | Senator Paul R Feeney | abstained |
| 6 | Michael R. Green, M.D., Ph.D. | Not present |
| 7 | Julie D. Gortze, RN | yes |
| 8 | Guadalupe Hayes-Mota, MBA, MS, MPA | yes |
| 9 | Lena Joseph, RN, CPN | yes |
| 10 | Representative Hannah Kane | yes |
| 11 | Andrew A. Lane, MD, PhD | yes |
| 12 | Representative Jay Livingstone | yes |
| 13 | Jeff R. Livingstone, PhD | yes |
| 14 | Diane Lucente, MS, LCGC | yes |
| 15 | Alexsandra B. Mahady | yes |
| 16 | Jenn McNary | yes |
| 17 | David T. Miller, MD, PhD | no |
| 18 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr) | yes |
| 19 | Shivang Patel, Pharm.D. | yes |
| 20 | Asma Rashid, MS, CGC | Not present |

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| --- | --- | --- |
| 21 | Michele Rhee, MBA, MPH | Not present |
| 22 | Robert E. Schultz, MBA | Not present |
| 23 | Celia Segel, MPP (Kate Mills) | abstained |
| 24 | Michael Sherman, MD | yes |
| 25 | Glenda E. Thomas | Not present |
| 26 | Ryan Thompson, MD, MPH | yes |
| 27 | Dylan Tierney, MD, MPH | yes |
| 28 | Ann Wessel, MS, RD, LDN | yes |
| 29 | Ross Zafonte, DO | yes |

**D Tierney** introduced the workgroup updates. He asked for a report from each workgroup.

**Workgroup 1 – presented by Tai Pasquini**

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

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

**T Pasquini** stated that workgroup 1 had met once in February but they had divided up a lot of work for people to work on between meetings. She stated that after much discussion, the group decided to write a white paper to address the question of prevalence. They were still in the process of finalizing the white paper agenda but we have more information after the next meeting. The group decided to look at common rare diseases that have reliable data sources. We plan to use the state and national data we find to validate other national data reports on rare disease prevalence. This way we can validate if MA is in line with the national figures or if we need to look at things differently. We also discussed that we need to include burden of disease in our white paper. We discussed the possibility of including stories to address the burden of disease.

She went on to state that the group decided to divide up the most common rare diseases that they felt had good data sources. Each of us took a different disease and then we will review that data when we meet in April. The disease we are looking at are hemophilia, Ehlers–Danlos syndromes (EDS), spinal muscular atrophy (SMA), Duchenne muscular dystrophy (DMD), cystic fibrosis (CF), and sickle cell disease. We chose these because we felt that these diseases had good data both nationally and state specific, but if anyone knows of another rare disease that may have equally reliable data sets, please let me know. She stated that one person that is no longer on the committee was looking into CF.

**L Joseph** volunteered to look into CF data.

**T Pasquini** asked the rest of her group if anyone wanted to share the work they were doing.

**A Dwyer** shared that he was looking into EDS. His research showed that there was no public dataset for EDS in MA. Some nordic countries have good data. He also stated that EDS specifically might have some data issues due to miscoding. He stated that this is very common with rare diseases because there are not often codes to match the diagnosis.

**A Lane** added that he had contacted the FDA office of rare diseases and discovered that they do not collect or store any prevalence data on rare diseases. They rely on their submitters to send data with their proposals. He did get information from NIH. They have a few data sets that could be helpful and ones we can use for validation. Sickle cell specifically has decent data due to newborn screening. MA does not share its newborn screening data, so there is no MA-specific data.

**A Wessel** shared that she researched hemophilia. CDC actually posts prevalence data for hemophilia. The data seems pretty good. I checked with Rich Pizzello, the Executive Director of the New England Hemophilia Association, and he thought the number was a little off. He felt that hemophilia was undercounted.

**T Pasquini** Thanked Ann and Andy for their feedback. She stated that in her work, she has noticed that many rare disease advocacy groups feel that their prevalence data is undercounted as well. She also wanted to let everyone know that the chair, David Miller was at a conference, and that was why she was reporting. David is looking into DMD and SMA.

**D Tierney** thanked Tai and workgroup 1 for all their hard work. He asked if the group was planning to present one number for prevalence of rare disease in MA or where they going to list the specific diseases.

**T Pasquini** stated that they planned to report one number but would use the disease-specific data to validate the numbers they got. We may end up using ranges or age-specific data, but we need to discuss that more at the next meeting. She then asked if anyone knows of any other disease data sets that we could use, please reach out. Sometimes, centers of excellence have really good data for specific diseases because they end up seeing 90% of the patients with that specific disease.

**J McNary** asked if they looked at other organizations that collect prevalence data? Is it worth surveying some of these groups to see what kind of data they collect.

**T Pasquini** responded by saying that it was a good idea. They hadn’t really thought about that for prevalence data. They had discussed reaching out to other groups for the burden of disease data. We know about the “Every Life Report” by the Every Life Foundation. Maybe we should reach out to them to see what kind of data they collect and would be willing to share.

**D Tierney** asked how they planned to measure burden of disease. Would you use adjusted life years or something like that?

**T Pasquini** stated that the group hadn’t really discussed how they would calculate the disease burden. We plan to discuss that at our next meeting. We do know there is data related to the quality of life, the every life report, and other reports on opportunity cost and hard cost, but we need to discuss this more. We do know that this could be a separate project in and of itself, so I think that the only thing we can do in this white paper is exemplary stories. I don’t think we will really have time to dive in much deeper than that.

**Jeff Livingstone** added that he remembered a presentation from NORD that calculated a more accurate accounting of rare diseases by state, but he didn’t remember specifically. He suggested asking NORD about this. He also suggested that the Rare Disease Diversity Coalition (RDDC) may have some data.

**A Reed** from NORD responded by saying that NORD does not have state-specific data but is looking into it. Harvard is now involved in helping them with state-specific data, so we are hoping to be able to help states with those numbers.

**D Tierney** thanked all for the robust discussion. Workgroup 1 is making great progress.

I’d like to now introduce a report for workgroup 2. He stated that Workgroup 2 is chaired by Jenn McNary. Their charge is to *advocate for and improve access to social supports and services for people impacted by rare diseases.*

With a goal of developing a profile of rare disease social supports and services in Massachusetts. I’d like to introduce Jenn.

**J McNary** stated that the group had met once. The first thing we did was verify that the charges were all charges that we felt were in our workgroup’s scope of work. All agreed.

We also agreed that there might be overlaps and cross-overs with the other groups. We would like to work together whenever possible, so we don’t duplicate work.

We think the first thing would be to develop a comprehensive list of resources and social supports. It may sound easy, but we think it will be very challenging. Once we compile this list, we want to survey the resources to better understand the resource and how it’s accessed. We need to know how hard or easy it is to access the services. We also want to survey the public to get their thoughts. We want to hear from them about what resources they use and what resources they can’t find or don’t have access to.

I can give you an example:

For those who don’t know, I have two sons with DMD. My older son is 24 and lives on his own. He was recently hospitalized, and we discovered he qualified for a visiting nurse to come to the house every day. The sad part is that he has qualified for this for ten years, but we never knew. How is it possible that we never knew this?

Anyway, we are starting with compiling all the resources we can find. Audrey from Representative Livingstone’s office has set up a google doc and has offered to help compile and organize the data. Lisa Deck has also offered to help.

We will send out the link to our google docs, and we would be grateful if you filled in anything you know of.

Google Doc for social supports and resources: <https://drive.google.com/drive/u/0/folders/1hlDocvgDN5VyapgZIqi-lJO7AC_hrmec>

**J McNary** stated that the next activity of the group would be to survey the resources to gather more information. At our next meeting, we will be working on developing the survey. If any other groups want to add questions to our survey, it may be a good way for us to collaborate. She also added that the group would like to figure out a way to elicit information from the public. We want to make sure we don’t miss anything either about a resource that exists or one that is needed and not available.

**R Thompson** suggested working with some of the hospital case managers and discharge planners. These people are paid to help our patients. They may know of some resources but may also be able to let you know what resources they struggle to find for patients and families. This could be a secondary audience that would really benefit from your work.

**J McNary** stated that it was a great point and thenasked if he knew the best way to reach these people. Also, what would be the best way to elicit information from them.

**R Thompson** offered to connect Jenn to the hospital-based case managers and discharge planners within the MGH-Brigham system. I’m sure a good first step would be to talk with them.

**J McNary** then suggested that it might be a good idea to ask what kind of tool would be helpful. Would it be a booklet, online database etc.

**J Gortze** we are Rare New England and know how valuable resources are for patients and families with a rare disease. I wondered if you looked into the Federation for Children with Special Health Care Needs Family Ties of Massachusetts. They may have some thoughts. I know that doctors often don’t even know where to find resources. They are often so busy treating the disease they don’t know where to find the supports that the patient needs.

**J McNary** stated that it sounds like we need a marketing plan. We need to figure out a way to get thee word out about the resources we find.

**A Mahady** stated that family ties have an online database, and they have parent coordinators to help manage what you are looking for.

**T Pasquini** added that she knows that sometimes, even within a hospital system, we don’t know what is available. There can be inconsistencies. I also think we need to look into MassHealth. There are resources within MassHealth that are hard to find or hard to access. Maybe insurance plans too.

**J McNary** stated that Tai brings up a really good point. Payers. We probably need to look at insurance, including MassHealth. I know that sometimes a resource is available, but insurance won’t pay for it. We need to identify what resources are available and how accessible they are. I think we also need to look at payers. I look forward to making recommendations about that.

**D Tierney** you said something that is really important. Just to re-emphasize, our job is to make recommendations to the legislature and those who have the power to make changes. We can’t do the work ourselves. We need help, and the best way to do that is to give a detailed recommendation to the legislature.

**J McNary** stated that she was very excited that we have the ability to make recommendations.

**D Tierney** thanked Jenn and Workgroup 2 for their work. He looks forward to seeing their recommendations. He then introduced Workgroup 3.

Workgroup 3 is chaired by Lena Joseph and their charge is to *Foster communication and collaboration to empower the rare disease community in Massachusetts.*

Their goal is to develop a profile of rare disease expert individuals, community-based organizations, voluntary organizations, healthcare providers, and any other public or private organizations with interest in rare diseases in Massachusetts.

**L Joseph** let everyone know that the workgroup had met once in February. They discussed their actual charge and how they would get it done. The group decided that the first step should be to create a comprehensive list of stakeholders. Although it seems fundamental, we felt that this would be our first step. We decided to split up the work and create a google doc that everyone could add to. We created a google doc (below) and added stakeholder groups. The groups will we considered as stakeholders are: legislators, hospitals, dieticians, nurse groups, doctor groups, pharmacies, industry, geneticists, health plans, academic institutions, research institutions, caregivers, biotech, and entrepreneurs. If anyone has any suggestions or any that we missed, please let us know.

**D Tierney** asked if the group had a goal to make recommendations. Creating a database requires maintenance. Maybe the recommendation is to find an organization to host and maintain this type of database.

**L Joseph** thanked D Tierney for that comment. We didn’t really think about that point, but you are right; maintaining something like this would be huge. We didn’t really think about sustainability.

**J McNary** also noted that workgroup 2 database would require the same kind of upkeep and maintenance. Maybe it would make send to recommend that we find someone or an organization to maintain it and fund a person. It will be so important to have someone to help people manage the information. I think it makes sense for us to work together on this.

**D Tierney** stated that he was struck by Lena’s report and thinking about not re-creating the wheel. I reached out to NORD to see if they may have some resources or information that will help our workgroups in meeting their goals. I asked NORD to come back and present information about resources from NORD that may help us. I’d like to introduce, Annissa Reed, the state policy manager for the eastern region from NORD to present information for us to consider.

**A Reed** first presented an overview of NORD. NORD was created in 1983. She then talked about NORD’s relationship with the FDA. She stated that they have a diverse network of partners and members. The organization has been in existence for 40 years. We are an umbrella organization with members that are 501(c)3 organizations. Many are organizations that advocate for a specific rare disease. We work with them to support them and help them connect with other organizations. We know of 1200 advocacy groups. We also work with advocacy groups that want to start an advocacy organization.

Our membership teams can query these groups by state so that may be helpful for your workgroups.

NORD has a rare disease database that provides information for patients and providers in non-technical language. There are interactive videos. We constantly strive to update this information. NORD also provides several 1-pagers to summarize a specific rare disease.

NORD created a new initiative, our centers of excellence. Researchers work with patients and doctors to interconnect centers in 23 different states. Harvard is one of our centers and is in Massachusetts.

Another incredible resource is our patient assistance programs. Since 1987 we have provided lifesaving support for many different patient services.

Our communication center is available for patients and providers can access our information.

NORD also has a state report card. This is a place to look at states and evaluate how they can support those with rare diseases. It is just a tool Some states have found this tool helpful for developing policies.

NORD also created advocacy and policy task forces. These task forces are set up to help advocacy groups come together to work on policy and collaboration.

NORD is always working to add new RDACs. We help with state-based coalitions. We encourage RDACS to work with each other and will help whenever we can help this collaboration.

We develop toolkits and supports for RDAC, and we would like to work with MA if there is anything we can do to support your work.

**D Tierney** thanked Annissa for your presentation and the work that NORD does to support RDACs nationwide. He asked a question about how NORD collects data on stakeholders. How do you collect and maintain this information?

**A Reed** stated that NORD has a whole department that maintains the stakeholder database. Maybe one way to share information would be to let you know when any MA-based organization is holding an event.

**D Tierney** asked if the stakeholder database was a comprehensive list of list of only strong, reliable organizations.

**A Reed** stated that her members pay a fee to be part of the stakeholder partners.

**D Tierney** thanked Annissa and NORD for the work they do and for sharing all this information with us. He then went on to ask if there were any announcements.

He gave an update on Rare Disease Day. Stating that Rare Disease Day was canceled because the state house closed on February 28th for a potential snow event.

MassBio has reimagined the event and is now holding a panel discussion on March 20th. They are asking people to register if you want to view it in person or online. They will also record it for later viewing if you cannot attend. You can register at MassBio.org if you are interested.

He also let the group know that the steering committee will be discussing a tool to collect contact information on our website from people who may be interested in learning more about the work we do.

The last question he posed to the group was whether or not they would like to try to hold an in-person full council meeting. If so, what do you think about May?

**J Gortze** stated that she thought it would be a good idea to have a hybrid version for those that may not be able to attend in person.

**J McNary** thought that May was too quick and the summer may be difficult, so could we look to September for this type of event?

**D Tierney** stated that Mary Lou would contact people about this, but we would look toward September and work on a central location for all. He then asked if there were any other announcements.

No one responded so he asked if there as a motion to adjourn.

**M Sherman** made a motion to adjourn

**T Pasquini** made a second to the motion

**D Tierney** asked if all were in favor of adjourning. All agreed

The meeting was adjourned at 10:43 and stated that the next full council meeting was May 18