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

**MEETING MINUTES**

REMOTE MEETING:    Thursday, July 20, 2023, 9:00-11:00 AM

*APPROVED September 21, 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.**

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

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

**D Tierney** reviewedthe agenda and introduced **Dr. Ryan Thompson**, a member of the council who would be facilitating a provider panel on the challenges of managing a complex patient in a rural setting.

**R Thompson** introduced the segment by talking about how challenging it has been to manage a patient who has a rare disease with these two panel members. He let all know that these providers go over and above to manage this patient and it has been a huge challenge for all, including the patient. He thought it would be helpful for all to hear directly from them about some of the challenges of managing complex patients in rural Massachusetts. He asked all to think about recommendations that the council could make, that would help providers like our panel members. He asked each of the providers to introduce themselves.

**Dr.** **Gregory Braden** introduced himself as a nephrology specialist who practices in W Springfield. He cares for many complex patients and finds it more challenging every day. Although he is a specialist, he often provides primary care to many of his patients because there is a lack of providers willing or able to manage complex patients. He provides care at Baystate Medical Center.

**Dr. Hanno Muellner** introduced himself as a primary care provider in Williamsburg Massachusetts. He provides care at Cooley Dickinson Hospital, which is now a part of the Massachusetts General Brigham network.

**R Thompson** gave a summary of the patient but explained that this is just an example and it could be any complex patient. He let all know that he became involved because the rural setting wasn’t always able to provide the care and supports needed to manage her care. He asked the panelists to share some of the challenges that they see in managing complex patients in a community setting.

**Dr. Mueller** began the discussion by stating that he felt that one of the most challenging part was the lack of continuity of care. He stated that the patient has many providers and some are in different healthcare systems and use different medical record systems, therefore he cannot see all the information needed to manage her care. He stated that he often has to ask the patient help.

**Dr. Braden** stated that he felt that one of the biggest challenges was the lack of specialty providers in the western part of the state. He stated that the financial reimbursement for providers in Western Massachusetts was far less than in the rest of the state. Providers can move toward the eastern part of the state and make more money so they leave. It is very challenging getting providers to stay in this part of that state. The other big issue that is related is the reimbursement rate. With the lowest reimbursement rate in the state, providers are not able to provide the care that is needed. He stated that without a connection to the Massachusetts General Hospital (MGH) Complex Care unit, he would not be able to provide the care that he does. He feels that MGH has his back. He also feels that many providers in Western Massachusetts, may not even know about this unit at Massachusetts General Hospital. He and his practice, have a strong belief that ALL patients should receive the same care, regardless of income but he finds it very challenging to do this. Consequently, we provide services that are not reimbursed or reimbursed at a rate that doesn’t even cover the cost of the service. Dr. Braden talked about young providers and their work ethic. Some of the young providers I have met are not willing to provide a service that isn’t reimbursed or not willing to put in the hours required to manage a complex patient.

**Dr. Muellner** commented that we needed more education for rural providers about rare diseases and managing complex patients.

**R Thompson** asked if there may be opportunities to support rural providers through a project like ECHO? He asked Dr. Muellner if he could think of any solutions that would help him manage complex patients in Western Massachusetts.

**Dr. Muellner** discussed that the fragmented communication between existing EMR systems was one of the biggest challenges that he faced. If there were a way to see medical information from all the providers that manage a patient, it would really help. Because of the lack of specialists in Western Massachusetts, patients often have to travel a distance and go to different medical systems.

**R Thompson** asked the council membership if they had any questions for the panelists.

**A Mahady** stated that she knew that there was not silver bullet but she wondered if the panelists thoughts that telehealth could help.

**Dr. Braden** stated that during COVID they were allowed to bill for telehealth visits and it was hugely helpful but since COVID policies ended, they have not been reimbursed for any telehealth visits. This would be so helpful if we could bill for telehealth, especially as some of our patients have difficulty getting to an in-person visit.

**T Pasquini** stated that as a resident of E Longmeadow, she knows first-hand about the physician shortages in western Massachusetts, especially for specialty care. She asked the panel what they think can be done about this.

**Dr. Braden** stated that 80% of the care his practice provides is for Medicare or Medicaid patients. The reimbursements for public programs are significantly less than private insurance reimbursement. He stated that he wasn’t sure how things would change unless the reimbursement rates were increased. He stated that they can’t balance bill for anything if it’s a Medicare or Medicaid patient. Currently, his practice needs to pay for a full-time person to do billing and another full-time person to manage insurance appeals. We know there are huge disparities in care between the eastern part of the state and the western part, but providers can’t make it work financially to practice out west.

**C Segel** stated that the Health Policy Commission looks at disparities based on geography. She asked the panel if they thought the disparities were more related to the financial reimbursement.

**Dr Braden** stated that about 90% of the care at Baystate was low-income and most are on Medicare or Medicaid. With reimbursements so low, it creates lots of challenges.

**A Dwyer** Thanked Dr. Muellner and Dr. Braden for their time. He thanked them for the great service they are providing to the residents of Western Massachusetts. He thanked them for outlining the issues they face in caring for patients every day. He asked the panel if they thought that AI or other systems could be used to help.

**Dr. Braden** stated that he thought that AI would be helpful in the future, but it’s not there yet. He felt that AI was only as good as the data and the people who entered the data. It may be a helpful tool in assisting with rare disease diagnosis, but there isn’t enough data yet.

**C Boney** stated that as the Chief of Pediatrics at Baystate, she could confirm what Dr. Braden said. Approximately 72% of the pediatric patients she sees are on Medicaid. She thought that provider training might be helpful and emphasizing team-based care could also be good. She stated that the silver lining of the pandemic was telehealth. We were able to do a lot of team-based care using telehealth. She agreed that we need telehealth for team-based care to be successful.

**R Thompson** Thanked the providers for their time and for the work they do caring for complex patients. He stated that we are looking for bridges and connections to better service our doctors and specialists, and we will hear shortly from Project ECHO. It’s one possible solution to help improve the care of rare disease patients. It will be interesting to see if it may be a tool we could use in Massachusetts.

**D Tierney** Thanked the panelists and welcomed them to join the Project ECHO presentation if they could. He then introduced Project ECHO presenters Ken Fraser and Elisa Buteau.

**K Fraser** introduced himself as the outreach director of Project ECHO (The presentation is in the meeting materials). He stated that Project ECHO is about building equity in outcomes. He stated that the project stated was about having the right knowledge in the right place at the right time. The benefits of knowledge can empower communities to access expert knowledge to save lives.

He stated that Dr. Sanjeev Arora founded Project ECHO because of a patient he saw. She was 43 years old and had Hepatitis C. She didn’t seek treatment because she lived too far from a place where she could get treatment. She died, and it was preventable. That’s why Dr. Arora created the ECHO Model as an “all teach, all learn” approach to bringing knowledge to rural and underserved communities.

Ken Shared slides with the council summarizing the Project.

He stated that Project ECHO has looked at how it could support rare diseases, and if Massachusetts is interested, they would be happy to discuss further. He stated that care that requires guided practice is when ECHO is most effective. If webinars are effective, then that should be the tool that you use. If guided practice is needed, then ECHO may be helpful.

For example, when learning to drive a car, it may not be effective to watch a webinar to learn to drive a car. It needs guided practice.

**D Tierney** Thanked Ken and Project ECHO for sharing this information. He then asked the council if they thought that Project ECHO could be used as a tool to help manage complex patients in communities. He added that ECHO could be used to build expertise in caring for a specific disease, but does the group think it can be used more broadly as sometimes there is only one patient with a specific rare disease?

 **J McNary** Systems are great in supporting doctors, but it seems that our healthcare system is already over-stressed. I can’t see how providers will find time to participate in this kind of learning tool. She stated that she didn’t think our providers have the bandwidth to do this kind of project.

**D Tierney** Could it be a way to centralize care?

**J McNary** stated that she has a great PCP for her son and doesn’t see how he would have time to log into something like this.

**K Fraser** added that ECHO recognizes this concern, but ECHO has found that once a provider logs on and gains the knowledge, they are excited to participate. We track their participation and have seen that once they get onto the system, they keep coming back.

**E Buteau** added that ECHO has seen providers see the possibility of collective discussion, especially with a difficult diagnosis. Working together sometimes allows them to get to a diagnosis faster.

**D Tierney** asked if this tool could be used for getting to an expedited diagnosis.

He added that the purpose of this presentation was to open up a discussion about tools to help the providers who care for patients with a rare disease. Does the council think that Project ECHO could be used as a tool for caring for patients with a rare disease?

No one from the council had anything to add.

**D Tierney** stated that we would now shift to workgroup 1 report on prevalence.

**D Miller (chair of workgroup 1)** first thanked the workgroup for its hard work on the prevalence white paper. He summarized the discussion by saying that it was hard to get accurate numbers, especially as some of the rare diseases are not even coded. There are a few diseases that make up the majority of rare disease prevalence. We took the approach of using the prevalence estimates for those few rare diseases and then validated those estimates against population data. We think we have presented a realistic estimate of the prevalence of rare diseases in Massachusetts.

He added that the draft was sent to the whole council for review. The workgroup is hoping that any council members who would like to comment, make edit suggestions, or provide any feedback would do so by responding on the Google document link that Mary Lou sent. Our workgroup will review all edits and comments and decide the most appropriate revisions. We will review all feedback prior to our next meeting on Monday, August 7th, so we ask that any feedback be sent by July 31st. Our plan is to present the white paper at the next full council meeting in September.

**D Tierney** stated that he read it and thought it was great. He stated that we all recognize that there is no way to come up with an exact number and that we need to use an estimate. He added that he hoped that the group could state the definitive range because it will be referred to on a regular basis. He stated that he saw a visual example of presenting an estimate and shared the diagram with the council. (slide in the presentation).

**T Pasquini and D Miller** both shared that they really liked the visual example, and they would try to use it in the paper.

**Rep Kane** stated that she thought the report was great. She added that she liked the section on considerations for Massachusetts and asked if it was possible to compare our numbers with other states. She asked if the workgroup looked at other state data. She then asked if the group planned to talk about the number of people that were impacted by a rare disease.

**T Pasquini** stated that Orphanet data gives overall distribution. She stated that the workgroup looked to see if any other state had done this work, and they couldn’t find any. She believes we are the only state that has tried to define the actual rare disease estimates for their state. She stated that she would reach back out to NORD to see if there was any new information.

**D Tierney** added that other states have tried to get to their specific state estimates by conducting surveys, which have not proven very productive. We think that our work will be used as an example for other states.

**Rep Kane** stated that she thought it might be helpful to see if we could estimate the number over time. For example, could we estimate what the number was in 1980 and what the number was in 2000 or something like that? I think it would be important to show that the numbers are growing and will continue to grow.

**A Lane** asked if we should use the word disease or disorder? NORD (National Organization for Rare Disorders) uses Disorder not Disease so I was wondering what everyone thought.

**Rep Kane** stated that as one of the writers of this legislation, I think it would be fine to use disorder instead of disease as long as there was an explanation.

**D Tierney** stated that he thought that disease is more stigmatizing than disorder.

**T Pasquini** stated that she could explain why disorder was used. In 1983, it was the height of the AIDS epidemic, and it was thought that disorder was a more acceptable term. I think that’s all it was about.

**Rep Kane** stated that she felt either could be used as long as it was explained.

**T Pasquini** stated that she wondered if the word disorder was used until there was a definitive diagnosis. Sometimes it takes a long time to get a diagnosis.

**D Miller** stated that for the purpose of the prevalence report, he thought that we should state in the report that no other state has done this work.

**D Tierney** stated that we would reach out to NORD to get a definitive answer.

**D Miller** stated that Workgroup 1 planned to complete the review and present the paper to the full council in September for a vote.

**D Tierney** asked how the group envisioned getting the report to the legislature. Should we have a legislative breakfast or something like that to present it?

**Rep Kane** stated that she thought we should have a special legislative session to present the paper. She thought it would be a good focus for a meeting.

**D Tierney** asked if there were any announcements. No one responded.

He then stated that Workgroups 2 and 3 were working on a survey and were still working on finalizing it.

He then asked if anyone had any thoughts about holding a public town hall-style meeting in October. We are thinking that these meetings would be centered around collecting testimonials from the community. No one commented.

He then stated that we need to start thinking about our recommendations. We should think about the most pressing recommendations that we should present. We may want to think about presenting one or two recommendations per year.

Our next full council meeting is September 21st. We are hoping to have the writer of the Autism report to speak. We may want to use this report as a template for our report.

**G Thomas** asked if we should be tracking bills that may impact people with a rare disease?

**D Tierney** stated that he thought this was a great idea.

**G Thomas** stated that she would start working on a list.

**T Pasquini** stated that she was on the NORD public policy committee and she would check in with them about what they are tracking.

**D Tierney** asked if there was a process for tracking/responding to bills or legislation.

**C Trabilcy** stated that hearings usually occur between April and January. Testimonies are taken before the bills are acted on.

**D Tierney** conducted a roll call vote to approve the minutes from the last full council meeting on May 18th, 2023. HE asked if all received the minutes. All agreed.

He then asked if there were any edits or corrections. No one responded. He then said he would conduct a roll call vote to ratify the minutes.

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|  | **Council Member** | **Approved** |
| 1 | Charlotte M. Boney, M.D | - |
| 2 |  Janis Creedon  | - |
| 3 | Lisa Deck  | - |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | X |
| 5 | Senator Paul R Feeney (Maya Rabinovitz) | x |
| 7 |  Julie D. Gortze, RN  | - |
| 8 | Guadalupe Hayes-Mota, MBA, MS, MPA  | X |
| 9 | Lena Joseph, RN, CPN | - |
| 10 | Representative Hannah Kane  | X |
| 11 | Andrew A. Lane, MD, PhD | X |
| 12 | Representative Jay Livingstone (Cassidy Trabilcy) | - |
| 13 |  Jeff R. Livingstone, PhD | - |
| 14 | Diane Lucente, MS, LCGC | X |
| 15 | Alexsandra B. Mahady  | x |
| 16 | Jenn McNary | - |
| 17 | David T. Miller, MD, PhD | x |
| 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 | - |
| 22 | Robert E. Schultz, MBA | x |
| 23 | Celia Segel, MPP  | x |
| 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** stated that the vote passed, and the minutes were approved as stated.

He stated that it was coming up on 11:00 am. Was there a motion to adjourn?

**D Miller** made a motion to adjourn

**D Lucente** seconded the motion

**D Tierney adjourned the meeting at 11:00 am**