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

Thursday, March 28, 2024, 9:00-11:00 AM

Meeting Minutes Approved 5.16.24

**Welcome**

**Dr. Dylan Tierney**, council chair, welcomed everyone to the meeting. He informed everyone that it was open to the public, but only those on the agenda would be allowed to speak.

He thanked Representative Hannah Kane for all her hard work on the council for the past three years. He let all know that Representative Joe McKenna, co-author of the bill that established the council with Representative Kane, would take her place on the council for the next three years. He welcomed Representative McKenna and asked if he would like to introduce himself to the council.

**Representative Joe McKenna** thanked everyone for their work on the council and expressed his gratitude for being able to help the council meet its goals. Rep. McKenna stated he has a personal reason for investing in the council's work. His family has been greatly impacted by the rare disease hemophilia. He has seen the great treatment progress from when his grandfather was treated to now watching his nephew’s treatment. He is excited to work with all.

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

|  |  |  |
| --- | --- | --- |
|  | **Council Member** | **Present** |
| 1 | Charlotte M. Boney, M.D | no |
| 2 |  Janis Creedon  | no |
| 3 | ~~Lisa Deck~~ (resigned 11/23) | NA |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | no |
| 5 | Senator Paul R Feeney (Maya Robinovitz) | present |
| 6 |  Julie D. Gortze, RN  | present |
| 7 | Guadalupe Hayes-Mota, MBA, MS, MPA  | present |
| 8 | Lena Joseph, RN, CPN | no |
| 9 | Representative Joe McKenna | present |
| 10 | Andrew A. Lane, MD, PhD | no |
| 11 | Representative Jay Livingstone  | present |
| 12 |  Jeff R. Livingstone, PhD | present |
| 13 | Diane Lucente, MS, LCGC | present |
| 14 | Alexsandra B. Mahady  | present |
| 15 | Jenn McNary | present |
| 16 | David T. Miller, MD, PhD | present |
| 17 | Tai Pasquini, PhD, MPA (Sen. Bruce Tarr)  | present |
| 18 | Shivang Patel, Pharm.D.  | present |
| 19 | Asma Rashid, MS, CGC  | present |
| 20 | Michele Rhee, MBA, MPH | no |
| 21 | Robert E. Schultz, MBA | present |
| 22 | Yue Huang, MS | present |
| 23 | Gail Ryan, PharmD | no |
| 24 | Glenda E. Thomas  | present |
| 25 | Ryan Thompson, MD, MPH | present |
| 26 | Dylan Tierney, MD, MPH | present |
| 27 | Ann Wessel, MS, RD, LDN  | present |
| 28 |  Ross Zafonte, DO | no |

**Quorum was established and the meeting was called to order at 9:08 am**

**Dr. Tierney** introduced guest speakers**: Sheilah Gauch and Jennifer Vitelli,** who are both parents of children with a rare disease. They are also both involved with the PANDAS PANS Advisory Council, which was established by the same legislation as the Rare Disease Advisory Council. He gave everyone the definition of PANDAS PANS, stating that

**PANS** is an abbreviation for Pediatric Acute-Onset Neuropsychiatric Syndrome. **PANDAS** is a subset of **PANS** and stands for **Pediatric Autoimmune Neuropsychiatric Syndrome Associated with Streptococcus.**

**PANS and PANDAS** are medical conditions brought on by post-infectious autoimmune encephalitis. This condition occurs when the body’s immune system mistakenly attacks healthy brain cells, leading to brain inflammation. In essence, it’s a misdirected immune response. Although the physiological mechanisms are the same, **PANS and PANDAS** are defined by different triggers.

He then introduced Sheilah.

Sheilah shared a slide presentation (attached with meeting minutes). She explained that the disease mostly impacts children but can also carry into adulthood. She talked about its impact on both the child and the whole family. Because of the variable symptoms, children are often diagnosed with mental illness, and medical intervention is often abandoned. She talked about the lack of awareness about the disorders, and the high number of misdiagnosed and underdiagnosed children.

She went on to discuss her journey raising two children with PANDAS. She stated that their presentation was very different. It was only through her persistence as a mom, to get them diagnosed. Once diagnosed and treated, they became different children. They are both in college now and doing well. This is one of the reasons she is so passionate about this work. She has seen the benefit of proper diagnosis and treatment. She thanked all the legislators for their support.

She then introduced Jennifer, who added that there was a need for more informed medical providers, especially in Massachusetts.

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

**J. McNary** asked if this was considered a rare disease.

**S Gauch** stated that as a mental health provider, she is sure that the system has missed many children. Without real data, it’s hard to know the actual number. For now, it is considered a rare disease.

**J Vitatelli** added that relentless parents have helped change the awareness and prognosis of these conditions.

**R Thompson** stated that he treats adults in the complex care division of Massachusetts General Hospital, and he wonders if some of his patients may be undiagnosed. He asked if there were any diagnostic tests.

 **S Gauch** stated that the diagnosis now does not have age restrictions. It was once considered only a pediatric disorder, but now we think there may be undiagnosed adults, so the age criteria for diagnosis have been removed. She added that Dr. Kyle Williams and Dr. Mark Pasternek from Massachusetts General Hospital wrote the guidelines for diagnosis, so they may be a good place to start.

 **J Vitatelli** added that she has been working on this effort for a long time but in 2019, we decided to bring our concerns to the people and are currently working on legislation that will improve the lives of those with PANS PANDAS.

**J McNary** thanked them both for their work and presentation.

**T Pasquini** also thanked them for their work. She asked how we can encourage people to follow their gut when facing obstacles to finding a diagnosis.

**J Vitatelli** stated that there was no internet when her child was exhibiting symptoms. There was no internet to look up information. There is a checklist that has been vetted and there are some recommendations for testing of inflammation markers. We can share this information if it’s helpful.

**S Gauch** stated that she is passionate because she has seen the impact of proper diagnosis and treatment. Medical must be included in the diagnostic phase. Otherwise, kids will be relegated to a life of treating symptoms instead of treating the root cause.

**D Tierney** thanked both Sheilah and Jennifer for their work and their time to speak to the council.

**D Tierney** moved to take a vote to accept the minutes from the last full council meeting on January 18, 2024. He asked if all had received the minutes. All acknowledged that they had. He asked if there were any edits or revisions. No one replied. He then asked for a motion to accept the minutes as presented.

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

**G Thomas** seconded the motion.

**A Roll Call vote was conducted by D Tierney.**

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

**D Tierney** stated that the meeting minutes were accepted.

He then explained that there would be a slight change in the order of the agenda. He introduced **Jenn McNary,** who will give an update on the community engagement workgroup.

Jenn explained that the workgroup met with two external marketing experts, who presented a marketing plan to increase the council's awareness. She stated that the workgroup would review this plan and present it to the steering committee. She said it was a small group, and they would welcome new members. She asked for any volunteers to reach out to her or put their names in the chat if they were interested in joining the group.

She also stated that she and Glenda Thomas were working on identifying legislation that may impact those with a rare disease, and if anyone would like to join them, they would welcome the help.

Members who volunteered to help included:

Rep. Joe McKenna, Rep. Jay Livingstone, Tai Pasquini, and Yue Huang.

**D Tierney** added that he had met with the DPH IT Department to see what could be done to update and modernize the website. He looked forward to working with the community engagement group to help make the website more user-friendly.

**D Tierney** then introduced guest speaker **Carolyn Sheridan.**

**Carolyn is the State Policy Manager at the National Organization of Rare Disorders (NORD).** Carolyn will talk to us today about the state report card for NORD.

**C Sheridan** shared slides with the council (attached in meeting materials). Carolyn first presented stats about NORD. The organization has been in existence for 41 years. She then reviewed the NORD report card. She presented the report card for Massachusetts, highlighting areas where Massachusetts has room for improvement. Areas for improvement include:

* Medical Nutrition
* Medicaid financial eligibility
* Newborn screening
* Telehealth services

**A Wessel** commented on medical nutrition, stating that in her experience, medical nutrition therapy is most often covered, but the co-pays and deductibles create a substantial hardship for her patients and families.

**Rep McKenna** added that he filed a bill to add Lysosomal Storage Disorders to the list of newborn screenings.

**D Tierney** added that newborn screening has been a topic of discussion in the past, and the council planned to have someone speak at the council soon.

**T Pasquini** asked Carolyn if adding a comment section to the report card would be possible. Stating that it may help clarify some of the details.

**C Sheridan** stated that a comment section was being considered.

**D Tierney** thanked Carolyn for presenting. He then gave an overview of the Legislative Briefing held on March 25th.

**The Legislative Briefing** was held on March 25 after being canceled in February due to a snowstorm. It was attended by legislators and their staff. It was the first legislative briefing of the council. We plan to have one next year at the same time.

One of the legislators' suggestions was to increase awareness of the council. One thought was to hold regional meetings to reach all geographic areas of the state.

**Rep McKenna** added that although it may be have been a small group, it was a great start and a successful 1st legislative briefing.

**Announcements—D Tierney** let everyone know that he and Rep Livingstone attended a rare disease event in Kendall Square in February. It was mostly attended by people in the life sciences industry. They gave a short overview of the council. Attendees were very interested and recommended that the council increase its exposure. It made me think about how to increase awareness and reach of the council.

**G Thomas** asked if it may be possible to hand out the 1-pager overview of the RDAC at a Myasthenia Gravis event.

**J Gortze** also asked about handing out the flyer at Rare NE events.

**D Tierney** stated that Mary Lou would send the 1-pager to all. Anyone could feel free to hand it out at any event they see appropriate.

**NEXT FULL COUNCIL MEETING: Scheduled for May 16, 2024.**

**ADJOURN: T Pasquini** made a motion to adjourn. **G Thomas** seconded.

**D Tierney adjourned the meeting at 10:53 am.**