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

REMOTE/VIRTUAL MEETING:    Thursday, June 23, 2022, 9:00-11:00 AM

APPROVED JULY 28, 2022

The meeting was called to order at 9:05 am by Dr. Dylan Tierney, Council Chair

Dr. Tierney welcomed two new people to the Council.

Introductions:

Lena Joseph – A new member of the council. Lena is a registered nurse who works at Boston Children’s Hospital in the cystic fibrosis clinic. The clinic works with patients with rare diseases, and she is very excited to be part of the council.

Mary Lou Woodford – the new council coordinator. Mary Lou is a registered nurse who has worked in many clinical, community, and public health settings. She was the Director of Women’s Health at the Massachusetts Department of Public Health for almost 20 years.

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

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|  | **Council Member** | **Present** |
| 1 | Charlotte M. Boney, M.D | X |
| 2 |  Janis Creedon  | no |
| 3 | Ms. Lisa Deck  | no |
| 4 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | X |
| 5 | Michael R. Green, M.D., Ph.D.  | no |
| 6 |  Julie D. Gortze  | no |
| 7 | Guadalupe Hayes-Mota  | X |
| 8 | Lena Joseph | X |
| 9 | Representative Hannah Kane  | 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 | no |
| 16 | David T. Miller, MD, PhD | X |
| 17 | Tai Pasquini, PhD, MPA  | X |
| 18 | Shivang Patel, Pharm.D.  | X |
| 19 | Asma Rashid, MS, CGC  | X |
| 20 | Michele Rhee, MBA, MPH | X |
| 21 | Robert E. Schultz | X |
| 22 | Celia Segel | X |
| 23 | Michael Sherman, MD | X |
| 24 | Senator Bruce Tarr | no |
| 25 | Ms. 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 | no |

A quorum was established. (21 members present, eight absent)

**VOTE to approve minutes from March 31, 2022, full council meeting.**

Motion to approve minutes made by Rep. Hannah Kane

Second by Jeff Livingstone

Dr. Tierney asked if there were any questions or edits to the minutes. Dr. Jeff Livingstone stated that his name was misspelled. The “e” was missing from his name. Dr. Tierney noted that the minutes would be revised to reflect the changes.

Roll Call Vote to accept minutes with the correction of “Livingstone” instead of Livingston

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|  | **Council Member** | **Vote Yes to accept** |
| 1 | Charlotte M. Boney, M.D | X |
| 2 | Andrew A. Dwyer, PhD, FNP-BC, FNAP, FAAN | X |
| 3 | Guadalupe Hayes-Mota  | X |
| 4 | Lena Joseph | X |
| 5 | Representative Hannah Kane  | X |
| 6 | Andrew A. Lane, MD, PhD | X |
| 7 |  Jeff R. Livingstone, PhD | X |
| 8 | Diane Lucente, MS, LCGC | X |
| 9 | Alexsandra B. Mahady  | X |
| 10 | David T. Miller, MD, PhD | X |
| 11 | Tai Pasquini, PhD, MPA  | X |
| 12 | Shivang Patel, Pharm.D.  | X |
| 13 | Asma Rashid, MS, CGC  | X |
| 14 | Michele Rhee, MBA, MPH | X |
| 15 | Robert E. Schultz | X |
| 16 | Celia Segel | X |
| 17 | Michael Sherman, MD | X |
| 18 | Ms. Glenda E. Thomas  | X |
| 19 | Ryan Thompson, MD, MPH | X |
| 20 | Dylan Tierney, MD, MPH | X |
| 21 | Ann Wessel, MS, RD, LDN  | X |

Dr. Tierney introduced **Michele Rhee**, a member of the council, a patient, and an advocate.

Michele gave a compelling summary of her journey to diagnosis, her challenges with treatment, and her experiences living with a rare disease. She talked about how her journey began almost 20 years ago. She was first diagnosed with thyroid cancer at age 19. Later she developed a benign tumor in her heart, then several tumors in her lungs. After years of tests and hospitalizations, she was finally diagnosed with Neoplastic Disorder. A rare disease that affects less than 800 people nationwide. Her diagnosis means that her body develops tumors randomly and spontaneously. She is constantly afraid that a tumor will present and she will need surgery. She expects to have at least one surgery per year. She knows that most people with her diagnosis are disabled, often bedridden, and cannot work. She feels blessed that she has a good job that pays well and that she comes from a very supportive family. She knows that many people with rare diseases do not have this. This is one of the reasons that she is so excited to be on the council. She wants to help in any way she can. She identified three priorities that she would like to see the council address.

1. People with a rare disease find it very difficult to hold a job. Without a job, health insurance is a problem. When people get a job, it’s hard to keep because of the ongoing medical appointments to manage their disease.
2. People with rare diseases often have multiple doctors and visit many different hospitals and medical institutions. Managing medical records is very challenging. Because many hospitals, doctors’ offices, and medical specialties do not share information, my responsibility is to manage my records. This is very challenging.
3. Coordinating my care is very exhausting and overwhelming. People with rare diseases often need help managing their care.

Question from Glenda Thomas;

How do you manage the cost of your care?

Michele: It is very challenging. I feel fortunate because I have been able to hold down a good job. I have been saving for my medical care since I first started working. I do not have children, so I keep every penny because I know I may need it for my medical expenses. The biggest challenge I see is the issue of “in-network” versus “out of network.” Visiting an ER can present a problem because even though the hospital may be in my coverage network, one of the doctors in the ER may be out of network. I always fear getting a surprise bill when I least expect it. Michele stated that she knows that in cancer patients, the financial toxicity often lasts much longer than the medical treatments.

Follow-up question from Glenda Thomas;

How do you manage post-surgical care?

Michele: It is challenging. I’m always exhausted. I am lucky and blessed, and I know it. I have saved for a long time and know that after surgery, I will have a lot of out-of-pocket expenses, but I have prepared for that. I know many people are not as fortunate as I am.

Comment from Dr. Michael Sherman

As an insurer, we are concerned about the surprise billing issue also. We want to help. I would like to know if there are gaps in insurance coverage that we as a council can address. I want to help and would be happy to talk with you to learn more.

Michele: Thank you. I will take you up on that. The biggest problem is the “in-network” and “out-of-network” issues. It’s very challenging for me to go on vacation. I have to research many things and call hospitals and ask specifically if they have doctors in my network and if the ER has “in-network” doctors to care for me if I need it. Unfortunately, I learned all this the hard way.

Dr. Tierney thanked Michele for her powerful and heartfelt story and thanked her for sharing.

Dr. Tierney talked about the challenges before the council. Most of the work has been focused on grounding the council and setting a foundation. The next step is to review the legislative language and set priorities. To help us think about the work ahead of us, we have three guest speakers from other RDACs to help us learn from them and guide us in developing our priorities and creating a plan. He asked that all questions be held until the three speakers have completed their presentations.

First to speak was **Tara Britt** from North Carolina. Tara is the president and founder of Rare Disease Innovations Institute, a nonprofit focused on educating, engaging, and equipping the rare disease community to achieve a higher quality of life, accelerating diagnosis, and enabling access and treatment through policy. She also serves as the Associate Chair of the North Carolina RDAC and Network. (Tara’s bio is in the meeting materials)

Tara has been a member of the North Caroling RDAC since its inception. North Carolina was the 1st RDAC in the nation, forming legislation in 2015. North Carolina has learned a lot since those early days. Sometimes we say that we wish we knew then what we know now. We hope you can learn from us as you begin your journey as a council. The most important thing would be to follow your legislative directive. Stay true to your legislation. You can’t be everything to all, so the best way forward is to follow directions from your legislature.

I would say the number one priority is newborn screening. At least it is for North Carolina. Identity and engage your stakeholders. Make sure you reach out to patients, biotech companies, foundations, patient advocate groups, academics, researchers, and clinicians.

Alabama and North Carolina have been around the longest and are going through some growing pains. We hope you will learn from us. One of the most critical issues we are now facing is membership on our council. It would help if you had clear rules for filling vacant seats or seats people no longer want. It can be very challenging if you need to rely on the legislature to replace a member. Make sure you have authority and the rules for replacing members.

I recommend setting three primary goals to achieve in your first year. (based on your legislation) You can’t do everything right away. As stated earlier, North Carolina’s #1 priority was newborn screening. We engaged a task force for this topic, and they wrote a white paper titled “Newborn Screening and Early Intervention for Treatment of Rare Diseases.”

Another recommendation is to stay visible with your legislature.

Tara also stated that she is working with NORD so that all RDACs can share resources. Sharing and learning from each other will help us all succeed. Tara thanked the council for inviting her and offered to help if the council needed it in the future.

Dr. Tierney introduced the next panel member.

**Dr. Nadia Falah** is an assistant professor at the Dept of Pediatrics, Division of Genetics and Metabolism at West Virginia University. She is a member of the West Virginia RDAC. (Nadia’s bio is in meeting materials)

Dr. Falah used a PowerPoint presentation, also included in the meeting materials.

Dr. Falah stated that the West Virginia RDAC was established through legislation in January 2020. It took about 12 months to seat the 12-member board. The first meeting was held in January of 2021.

One of the first activities of the RDAC was creating a website. One of the council members developed the West Virginia RDAC (<https://wvrare.org/>) website. The RDAC is supported by the West Virginia Department of Health and Human Resources.

The next activity of the council was to conduct a survey to determine the incidence and prevalence of rare diseases in West Virginia. NORD estimates that 250,000 people live with a rare disease in West Virginia. However, we have only had 72 responses to the survey so far. We have had a difficult time reaching people to complete the survey. We used the questions from Pennsylvania and modified them a bit. We created flyers to give out so that people would know where to go to complete the survey. We are not sure if the low response number is because the survey takes 30 minutes to complete or if we are just not reaching people. She reviewed specific questions and responses. (See her PPT for specific details) She thanked the council for the invitation and offered to help if needed in the future. She stated that she will be moving to North Carolina to do a fellowship at Duke University in the fall but would stay connected to the RDAC work.

Dr. Tierney introduced the third speaker**, Dr. Scott Strome**.

Dr. Strome is the Executive Dean and Vice Chancellor for Clinical Affairs at the University of Tennessee Health Science Center College of Medicine. He is also the chair of the Tennessee RDAC. He stated that he was eager to help Massachusetts in any way he could. He grew up in Massachusetts, went to Harvard Medical School, and still has strong ties to our state. (Scott’s bio is included in the meeting minutes)

Dr. Strome stated that we should consider the council’s connection to our state's medical schools and the well-respected hospitals we are so fortunate to have close by. The Tennessee RDAC utilized resources at the University of Tennessee, which was very helpful. Funding is a critical part of accomplishing the council’s goals. Most of the work done by the council will require funding. The University of Tennessee was able to assist with many of our activities.

Dr. Strome also stated that inviting patients to council meetings was a great way to keep the council reminded about the mission and help the council stay current with the daily issues facing people with rare diseases.

The next important thing to think about was identifying priorities for the council. In Tennessee, the #1 priority was access to genetic testing, and the #2 priority was newborn screening. He recommended that the council review the state and federal laws related to genetic testing and newborn screening to ensure that the laws and regulations are aligned. If not, that may be a priority for the council to work on. You may have to look at changing some laws or regulations in your state to align with the work the council wants to do. In Tennessee, the council wanted to work towards ensuring that all newborns born in the state would get genetic testing. There are challenges to this position. There are ethical issues, and we are still working on them. One point is in the interpretation of the data. Tennessee decided that it would only report on discernable mutations.

Another issue to think about is where would the data be stored? All of this is contingent on parental consent. To assist with this priority, Tennessee is investigating the work of Project Baby Bear, which provides whole genome testing. Tennessee plans to pilot this soon. He encouraged Massachusetts to look into the work of Dr. Stephen Kingsmore <https://radygenomics.org/2018/rady-childrens-launches-project-baby-bear/> on whole-genome testing for newborns.

Another activity we found very helpful was assigning a council member to priority stakeholder groups. That council member would be responsible for attending the stakeholder group meetings and reporting back to the council. This is a way to stay connected to what is happening in your rare disease communities.

Dr. Tierney asked if there were any questions for the panelists.

Michael Sherman commented that kids with rare diseases often spend more than seven years in their diagnostics journey. He asked Dr. Strome if insurers would be paying for the whole genome sequencing when roughly only 25% would lead to a diagnosis.

Dr. Strome stated that Tennessee has a largely rural area with little or no access to specialty care. One question Tennessee has grappled with is whether to ensure that all children get access to specialty care or whole-genome sequencing. They are still debating what direction will be best for Tennessee.

Andy Lane asked How can we work with academics? He stated that Massachusetts has a wealth of medical institutions and medical schools, and we should figure out how to tap into that. He suggested that the council engage with the Broad Institute, a well-respected Boston institution. He stated that he was on the board of the Broad and would be happy to make any connections. He also noted that the Broad Institute just convened a rare disease club so students can learn more about rare diseases and the challenges that patients face. He thought this was a great way to help students understand the complexities patients and families face when they have a rare disease. He also noted that Boston College just had a rare disease symposium to help students learn about the rare disease community.

Dr. Strome noted that Massachusetts has a large biotech community and several biotech companies have some connection to the rare disease community. Our council should investigate the biotech companies and what programs and services they may have for people with rare diseases.

Dr. Tierney thanked the panelists for their willingness to share their lessons learned. The panelists thanked the council for the invitation and said they would be willing to help whenever possible.

**Chairperson Updates**

***Steering Committee meeting held May 27, 2022***

Dr. Tierney stated that the RDAC Steering Committee met for the first time on May 27th. The committee discussed how the council should conduct its work. He also summarized the work of the council to date. The council has discussed how DPH can best support the council and has hired a coordinator. He suggested that the most effective way for the council to get work done may be to form working groups or subcommittees. He plans to discuss this with the steering committee during the next meeting on July 14th. He plans to have the steering committee review the legislative directives and develop a suggested list of workgroups or subcommittees to present to the full council on July 28th. Two possible workgroups for discussion may be a research group focusing on institutions researching rare diseases, clinical trials, etc. The other possible workgroup may be a promotion workgroup that would focus on increasing awareness of the council and its work.

He stated that the July 28th full council meeting would focus on the workgroup/subcommittee structure, begin drafting a mission statement, and start setting priorities for the council. The other priority for the council will be to create a set of operating procedures by which the council and subcommittees will work. The steering committee will also review our meeting schedule and draft a possible meeting schedule for the following year.

Dr. Tierney stated that the July 28th meeting would be very important.

He asked if there were any other questions, comments, or announcements from the membership. No one responded, so Dr. Tierney asked for a motion to adjourn.

Motion to adjourn: Michael Sherman

Second to adjourn: Lena Joseph

Meeting adjourned at 10:55 am.