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

**Remote meeting January 23, 2025**

**9:00 am – 11:00 am**

**Meeting Minutes – Approved March 27, 2025**

**Welcome:** Dr. Dylan Tierney, RDAC Chair, welcomed all to the meeting. He let all know that the meeting was open to the public but only those on the agenda would be allowed to speak.

He then introduced new council member, Kristen Shannon, LCGC who is replacing Diane Lucente LCGC. Kristen introduced herself to the council. She thanked all for the opportunity to join the council and was excited and passionate about this work. She stated that she was alicensed and certified genetic counselor at Massachusetts General Hospital. She has 28 years of experience working as a genetic counselor. She is a lifelong Massachusetts resident. Kristen would you like to introduce yourself to the council. She stated that she felt that access to good genetic counseling services was on ongoing problem. In rural areas it’s even worse than in larger urban areas. She hopes to be able to work on this issue.

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

|  |  |  |
| --- | --- | --- |
|  | **Member** | **Present** |
| 1 | Olaf Bodamer, MD, PhD, FACMG | X |
| 2 | Charlotte Boney, MD | resigned |
| 3 | Andrew A. Dwyer, PhD, FNP-BC | X |
| 4 | Christelle Moufawad El Achkar, MD | X |
| 5 | Senator Paul Feeney (Maya Rabinovitz) | X |
| 6 | Julie Gortze, RN | X |
| 7 | Heather Gray-Edwards DVM, PhD | Joined at 9:10 |
| 8 | Yue Huang, MS | X |
| 9 | Lena Joseph, RN | - |
| 10 | Representative Jay Livingstone (Cassidy Trabilcy) | X |
| 11 | Jeff Livingstone | - |
| 12 | Kristen Shannon, MS, LCGC | X |
| 13 | Alexsandra Mahady | - |
| 14 | Representative Joseph McKenna | X |
| 15 | Jenn McNary | X |
| 16 | Tai Pasquini, MPA, PhD | X |
| 17 | Shivang Patel, PharmD  | X |
| 18 | Asma Rashid MS CGC | X |
| 19 | Gail Ryan, PharmD | X |
| 20 | Robert Schultz | - |
| 21 | Glenda Thomas | X |
| 22 | Ryan Thompson, MD, MPH | X |
| 23 | Ann Wessel, MS, RD, LDN | X |
| 24 | Ross Zafonte, DO | - |
| 25 | Dr. Dylan Tierney, MD | X |

**D Tierney** asked if all received the minutes from the last meeting. All affirmed. He asked if there were any edits or corrections. No corrections suggested. He then asked for a motion to accept the minutes as presented. Rep McKenna made a motion to accept the minutes as presented. T Pasquini seconded. Roll call to accept minutes were as follows.

|  |  |  |
| --- | --- | --- |
|  | **Member** | **Accept** |
| 1 | Olaf Bodamer, MD, PhD, FACMG | X |
| 2 | Charlotte Boney, MD | resigned |
| 3 | Andrew A. Dwyer, PhD, FNP-BC | X |
| 4 | Christelle Moufawad El Achkar, MD | X |
| 5 | Senator Paul Feeney (Maya Rabinovitz) | X |
| 6 | Julie Gortze, RN | X |
| 7 | Heather Gray-Edwards DVM, PhD | X |
| 8 | Yue Huang, MS | X |
| 9 | Lena Joseph, RN | NA |
| 10 | Representative Jay Livingstone (Cassidy Trabilcy) | X |
| 11 | Jeff Livingstone | NA |
| 12 | Kristen Shannon, MS, LCGC | abstain |
| 13 | Alexsandra Mahady | - |
| 14 | Representative Joseph McKenna | X |
| 15 | Jenn McNary | X |
| 16 | Tai Pasquini, MPA, PhD | X |
| 17 | Shivang Patel, PharmD  | X |
| 18 | Asma Rashid MS CGC | X |
| 19 | Gail Ryan, PharmD | X |
| 20 | Robert Schultz | - |
| 21 | Glenda Thomas | X |
| 22 | Ryan Thompson, MD, MPH | - |
| 23 | Ann Wessel, MS, RD, LDN | X |
| 24 | Ross Zafonte, DO | - |
| 25 | Dr. Dylan Tierney, MD | X |

**D. Tierney** gave all an update from the last steering committee meeting. He let all know that the steering committee discussed priorities for 2025 and decided on the following.

* Medical Nutrition – continue the discussion
* Investigate and study the economic burden of rare disease in Massachusetts
* Develop a better understanding of the challenges related to the healthcare transitions between pediatric and adult care for those with a rare disease
* Investigate the possibility of developing a rare disease registry
* Improving access to resources and support services for those with a rare disease in MA

To address some of this, we have invited a speaker today to talk about the challenges of care transition between pediatric and adult care as well as the access issues for resources and services. He introduced guest speaker, Nathan Grant.

Nathan is the twin brother of Nik Grant, a 27-year-old adult who has a rare, neurodegenerative condition called MPS II (also known as Hunter syndrome). Nathan’s experiences with his brother have made him a passionate advocate in the rare disease community. Currently, Nathan is conducting research at Boston Children’s Hospital to identify strategies to help people with rare diseases transition effectively from pediatric to adult health care. Nathan is also the Founder and President of [Siblings with a Mission](https://urldefense.com/v3/__http%3A/www.siblingswithamission.org/__;!!CPANwP4y!SUTNz1tN4gKguiamKbioskLnQ5shdBqDqTSxb7cAy-z92HIMtw7F5O1Dk8yQfpd9EZm44SZKqlcpv5jKGZvfQ5hz3D8nRLgjawdkpA$), an international organization that supports siblings of people with complex health conditions and their families. From Cincinnati, Ohio, Nathan is currently a third-year MD/MBA student at Harvard Medical School and Harvard Business School. He received his undergraduate degree in biology from Harvard College and an MPhil in Health, Medicine, and Society from the University of Cambridge as a Harvard-UK Fellow. Inspired by his brother, Nathan hopes to help improve outcomes for people with rare diseases and their families through clinical care, research, and advocacy.

**N Grant** thanked everyone for the opportunity to speak to the council. He started by stating that his life and life mission is centered on the issues we are talking about. Nathan spoke comprehensively about the challenges faced by his brother and his family. He stated that his brother was diagnose at age 3 and when he was diagnosed, the doctors gave him a life expectancy of about 10-12 years. He is now 27 years old. As a result, he feels that his family was unprepared for the care required in adulthood. Nathan stated that his brother lives at home with his parents but needs periodic respite care to give his family a break. His brother is non-verbal and can have some serious behavioral issues, often violent. This has presented significant challenges finding caregivers and respite care. He stated that the challenges from pediatrics to adult care have been monumental. Some providers may continue to see his brother but other won’t or can’t. This creates significant challenges to coordinate his brother’s care. There is no consistency of who will continue to care for him and who will not. Nathan stated that finding a new adult provider has been beyond challenging. Most primary providers are not educate about his rare disease and not comfortable taking him on. Other are just too overworked and overwhelmed to take on such a complex patient, especially with behavioral issues. Nathan felt strongly that we needed to do a better job at educating doctors who care for adults, about rare diseases. He also stated that it would be really helpful to have help coordinating his brother’s care. His mom, who is not medically trained, is the one to coordinate all his care and as she ages, this is becoming more difficult. Nathan felt that help with coordinating care for complex patients like his brother, would be really helpful. He suggested a medical home for rare disease patients. The other issue he felt needed some discussion was related to equipment. For example, when his brother has to go for a procedure in an adult hospital, they often don’t have the equipment for his brother’s small stature. He felt that there needed to be more emphasis on ensuring the hospitals that treat adults should have size appropriate equipment. He also mentioned that pediatric hospitals often have behavioral support services when his brother needed a procedure. This really helped his brother’s and consequently his family’s anxiety. Most adult hospitals don’t have anything like this. The other major challenge for finding community based respite services. These faculties are often very expensive and hard to access. They spend an excessive amount of time finding, securing and maintain access to these programs and facilities. Appropriate community based services are few and far between. Nathan then thanked everyone for allowing him to share his story and was excited that the council was addressing this important topic.

**J McNary** asked Nathan if he could speak to the financial burden of doing all this and if there were caregiving issues he could specifically discuss.

**N Grant** stated that it takes a lot of time and resources to access and maintain many of the services needed for his brother and mom to have quality of life. He stated that they have had to engage lawyers and advocates to assist with this challenge. He added that when someone needs behavioral support like his brother, it’s even more difficult.

**O. Bodamer** asked if Nathan knew of any research being done on this topic.

**N Grant** He said that most of the research he had read was related to educating doctors and health systems about the care needed. He referred to some research that recommends a transitions of care sheet that can inform doctors, programs, and facilities about his brother’s condition and needs. He added that families of rare disease patients needed education in the early teen years about planning for the future. These conversations just don’t seem to happen.

**A Dwyer** added that he has done some research on this topic and found that there is a lack of community support, lack of access to community services, and a lack of coordination of care.

**N Grant** stated that there was also an added cost not often recognized. Caregivers, like his mom, cannot work because of the time commitment to support his brother. There is also the issue of caregiver burnout or even just times when a caregiver is sick or cannot work. He felt that this topic needed more research.

**C Achkar –** stated that she was at one of the top hospitals in the country and these issues are so prominent, she couldn’t image how hard it is for small or rural hospitals. She also stated that sometimes she continues to see patients with a rare disease even into adulthood, however, she can’t admit them to Children’s Hospital if they need to be admitted. She stated that it’s often hard for providers like herself as well. When we have to hand off a patients that ages out and into adulthood. She acknowledged that some of the conditions are so rare and she knows she understands the condition so well, it’s hard to transfer a patient to someone who has never treated the condition. She also validated that conversation about the aging out of pediatrics needs to happen early in adolescence.

**N Grant** added that families are often fearful about different areas of this transition also. We need to have conversations with families about what their fears are.

**D Tierney** thanked Nathan for his time and commitment to this topic. He invited him to stay for the rest of the meeting and join any future meetings.

He then introduced that next topic. Learning about resources and services available in Massachusetts.

He let everyone know that we had two guest speakers today Adding that we not only needed to understand the needs of those in the rare community, we needed to be able to identify the resources and services available and figure our any gaps that may exist.

He then introduced Eileen Davis from Mass211 and Call2Talk. Mass 211 is an easy to remember telephone number that connects callers to information about critical health and human services available in their community. It serves as a resource for finding government benefits and services, nonprofit organizations, support groups, volunteer opportunities, donation programs, and other local resources. Eileen Davis is the vice-president of Mass211 and she will give us an overview today.

**E Davis** thanked everyone for the invitation to speak and expressed willingness to work with or partner with the RDAC. She let all know that 211 was a state-wide call and referral center. The agents who answer the phone connect people to services. The call center agents are available 24/7 and have access to a translation line for over 100 languages. The database they use is constantly updated. There is a dedicated team focused solely on updating the database. She let all know that the biggest request received by the call center is for mental health an behavioral health resources. During the pandemic the center answered call related to COVID vaccine appointments.

She ended by stating that 211 would be open to partnering with the RDAC to make sure the agents have resource for the rare disease community. She also asked all members to review the 211 and if there are any resources not listed, they should send the info so that it can be added to the database. She thanked the council for the opportunity to meet the RDAC and is eager to learn more the rare disease community.

**D Tierney** then introduced Annette Peele from MassOptions.

MassOptions connects older adults, individuals with disabilities and their caregivers with agencies and organizations that can best meet their needs.

**Annette Peele** has worked in the older adult field for approximately 30+ years. Her responsibilities have led her to experience working in positions that assist older adults and people with disabilities connect to services directly as well as solidify policy and processes to ensure the programs and services exist and run smoothly with high quality. She is currently working at the Executive Office of Aging and Independence, formerly the Executive Office of Elder Affairs as the Director of Community Services.

**Annette** thanked all for the opportunity to speak to the council. Annette started by saying that the department is focused on aging and independence. Aging in the community is the focus now. MassOptions started in 2015 because the senior community wanted someone to help them get access to programs and services. MassOptions is a centralized resource for long term care and resources. As you talk about transitions from pediatric to adult care, she emphasized that the same issues often happen to older adults as they age. They often have to change doctors and it is very challenging for them.

Annette also added that they have a caregiver network. MassOptions provide resources for caregivers also. They partner with 211 and there is some overlap but the main difference is that MassOptions often make a warm handoff, while 211 mostly makes referrals or gives the information to the caller. The MassOptions staff will work with the caller to find services that they may be eligible for and often assist with the applications to the programs or services. She let the council know that the most requested services are for personal care assistants or for food and nutrition services.

**D Tierney** thanked Annette and Eileen for sharing this information with the council and asked them to participate in the discussion if possible. He added that the council have thought about creating a hotline as a way for the rare disease community to connect with services. He asked if anyone on the council had questions or thoughts on how these services may be helpful for the rare disease community.

**T Pasquini** stated that using what exists is the best way to go. She recommended that we evaluate the service that are currently available to see what gaps may exist.

**A Peele** added that MassOptions partners with other agencies.

**J McNary** asked who is eligible for their services. She added that she had two your men that were in wheelchairs and were told that they didn’t qualify.

**A Peele** responded by stating that her program has no eligibility criteria, they work with state programs and services and help people apply.

**E Davis** added that 211 is always looking to add information to their database. 211 relies on people letting them know what is available. She again encouraged anyone to notify 211 with resources that they think may be helpful for the community.

**D Tierney** asked if there was data to see who was calling 211 and how many were successfully connect to a program or service. He then asked if it would be possible to have a rare disease person to answer the calls for people seeking are disease resources.

**E Davis** stated that this could definitely be an option. She offered to have some of the call agents get trained specifically on rare disease issues.

**D Tierney** thanked all for the productive discussion and stated that we will take all this under advisement and discuss more.

He then asked the Policy committee for an update.

**Glenda Thomas** gave an update from the Policy Subcommitteefirst formal meeting on January 8th, 2025. She stated that the committee met on January 8th and formalized their mission statement and discussed next steps. She said that the committee will meet quarterly and if there was anyone else who was interested in joining the subcommittee they should reach out to one of them. They should contact Jen, Glenda or Mary Lou.

**D Tierney** stated that all subcommittees needed a set number of members because a quorum was required for all meetings, including subcommittee. He also added that the committees could invite other members, subject matter experts or others to attend their meetings.

**J McNary** asked if she could add some important information she just learned about. She spoke to

**D. Tierney** statedthat although there wasn’t much time, he asked if we could move forward in establishing two new subcommittees. He explained that there was a community engagement workgroup that met several times in the past year and asked to become a formal subcommittee. He asked if there was a motion to form a standing subcommittee to provide guidance and support for all community engagement activities of the RDAC by identify and organizing information about the rare disease community in Massachusetts.

**T Pasquini** made a motion to form the community engagement subcommittee. **A Dwyer** seconded. **D Tierney** then asked if all were in favor. (all agreed) he then asked if anyone was opposed. (no one opposed) He stated that the community engagement subcommittee was now a standing subcommittee of the council.

**D Tierney** then asked if there was a motion to form a research subcommittee. This committee would provide guidance and support for research activities of the RDAC by identifying and synthesizing existing research to provide evidence for RDAC initiatives including leading and supporting new research projects and collaborations. More specifically this committee would working on figuring out how we can study the economic burden of rare disease in Massachusetts among other research topics of the council. He asked for a motion to form this committee. **J McNary** made a motion to form a standing subcommittee called the research subcommittee. **T Pasquini** seconded. **D Tierney** asked who was in favor of forming the research subcommittee? (All responded “I”) He then asked if anyone was opposed. (no response) He then stated that the research subcommittee would be a formal standing subcommittee. The committee would be chaired by Tai Pasquini and if anyone was interested in joining the subcommittee could they reach out to Tai or Mary Lou.

**D Tierney** made a few announcements

**Rare Disease Day, February 28, 2025**

* **The Boston Globe presents Rare Disease Summit on February 25th, 2025 from 9:30 am – 2:05 pm**
* **MassBio Presents Rare Disease Forum on February 28, 2025 from 10:00 am – 1:00 pm**

**NEXT FULL COUNCIL MEETING would be on March 27, 2025 from 9:00 am –11:00 am.**

He then asked for a motion to adjourn

**A Dwyer** made a motion to adjourn

**T Pasquini** seconded

**D Tierney** adjourned the meeting at 10:57