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

**Remote meeting March 27, 2025**

**9:00 am – 11:00 am**

**Meeting Minutes – Approved 5.22.25**

**Dr Tierney Welcomed all to the meeting at 9:00.**

He then announced that the public is welcome to join any of the RDAC meetings. However, only guests were on the agenda would be allowed to speak during the meeting. For all guests on the call, he asked if they could please make sure that their video and audio were turned off.

He then stated that we had four new members join the council since our last meeting. He asked each of them to introduce themselves to the council.

He introduced:

Dr Stacey Cook, who is a pediatrician at Boston Children’s Hospital who administrates a structured clinical program for over one thousand children with rare diseases who have medically complex conditions and severe disabilities. Dr. Cook is replacing Dr. David Miller. He thanked Dr. Miller for his service to the council.

Dr. Eric Crombez, who is a board certified clinical geneticist, Chief Medical Officer and Executive Vice President for Ultragenyx where he oversees global clinical development and execution of the company’s gene therapy programs. Dr. Crombez is replacing Jeff Livingstone. He then thanked Jeff Livingstone for his work with the council.

Alison Griffin who is a registered dietician workking at New England Life Care. Her career has focused on helping those with complex conditions needing nutritional support. She has over 15 years of experience working in both the hospital and outpatient settings. Ms. Griffin is replacing Anne Wessel. He then thanked Anne Wessel for her work with the council.

Valarie Molyneaux who was diagnosed with Vitiligo in 2009. She is a mom and advocate for Vitiligo. Ms. Molyneaux is replacing Janis Creedon.

**D Tierney** then conducted a roll call to establish a quorum to start the meeting.

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|  | **Member** | **Present** |
| 1 | Olaf Bodamer, MD, PhD, FACMG | X |
| 2 | Stacey Cook, MD | X |
| 3 | Eric Crombez, MD | X |
| 4 | Andrew A. Dwyer, PhD, FNP-BC | X |
| 5 | Christelle Moufawad El Achkar, MD | X |
| 6 | Senator Paul Feeney (Maya Rabinovitz) | X |
| 7 | Julie Gortze, RN | X |
| 8 | Heather Gray-Edwards DVM, PhD | X |
| 9 | Alison Griffin, MS,RD,CSO,CNSC, LDN | X |
| 10 | Yue Huang, MS | X |
| 11 | Lena Joseph, RN | - |
| 12 | Representative Jay Livingstone (Cassidy Trabilcy) | X |
| 13 | Alexsandra Mahady | X |
| 14 | Representative Joseph McKenna | X |
| 15 | Jenn McNary | - |
| 16 | Valarie Molyneaux | X |
| 17 | Tai Pasquini, MPA, PhD | X |
| 18 | Shivang Patel, PharmD | X |
| 19 | Asma Rashid MS CGC | X |
| 20 | Gail Ryan, PharmD | X |
| 21 | Robert Schultz | X |
| 22 | Kristen Shannon, MS, LCGC | X |
| 23 | Glenda Thomas | X |
| 24 | Ryan Thompson, MD, MPH | X |
| 25 | Ross Zafonte, DO | - |
| 26 | Dylan Tierney, MD | X |

**D Tierney** stated that there was a quorum and he called the meeting to order at 9:19.

He then asked if all received the minutes from the last meeting and if there were any edits or revisions? No response so he asked if there was a motion to accept the minutes as presented.

Rep. Livingstone made motion to accept the minutes, T Pasquini seconded.

D Tierney conducted a roll call vote. He asked that if anyone was not present at the last meeting they should abstain.

**Roll Call vote was conducted.**

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|  | **Member** | **Approve** |
| 1 | Olaf Bodamer, MD, PhD, FACMG | X |
| 2 | Stacey Cook, MD | Abstain |
| 3 | Eric Crombez, MD | Abstain |
| 4 | Andrew A. Dwyer, PhD, FNP-BC | X |
| 5 | Christelle Moufawad El Achkar, MD | X |
| 6 | Senator Paul Feeney (Maya Rabinovitz) | Abstain |
| 7 | Julie Gortze, RN | X |
| 8 | Heather Gray-Edwards DVM, PhD | X |
| 9 | Alison Griffin, MS,RD,CSO,CNSC, LDN | Abstain |
| 10 | Yue Huang, MS | X |
| 11 | Lena Joseph, RN | - |
| 12 | Representative Jay Livingstone (Cassidy Trabilcy) | X |
| 13 | Alexsandra Mahady | X |
| 14 | Representative Joseph McKenna | X |
| 15 | Jenn McNary | - |
| 16 | Valarie Molyneaux | Abstain |
| 17 | Tai Pasquini, MPA, PhD | X |
| 18 | Shivang Patel, PharmD | X |
| 19 | Asma Rashid MS CGC | Abstain |
| 20 | Gail Ryan, PharmD | X |
| 21 | Robert Schultz | Abstain |
| 22 | Kristen Shannon, MS, LCGC | X |
| 23 | Glenda Thomas | X |
| 24 | Ryan Thompson, MD, MPH | X |
| 25 | Ross Zafonte, DO | - |
| 26 | Dylan Tierney, MD | x |

**D Tierney** stated that the minutes were approved.

Next he introduced our rare disease speaker, Caroline Walsh. Caroline is a 25 year old from Massachusetts who has a rare disease. She will speak to the council about her rare disease and the challenges presented when she had to transition from pediatric care to adult care.

**C Walsh** shared her story. She stated that she has a rare disease called Turner Syndrome, which is genetic condition that only affects females. It occurs when a female is missing all or part of the X chromosome(s). It can result in a wide range of symptoms that require regular follow-ups and testing. She expressed that care guidelines would be really helpful for doctors that aren’t as familiar with the diagnosis.

She stated that she began transitioning from pediatric care to adult care when she was about 20. She stated that it was really hard leaving her pediatrician who she had developed a strong bond with over the years. She doesn’t think there is enough emphasis on this. She then stated that another challenge of moving to adult care was the increased responsibility to manage her care herself. She felt that there were a lot more supports while under a pediatrician’s care.

She thanked the council for having her speak, and appreciated the work they were doing.

She then asked if there were any questions.

**D Tierney** asked how she found an adult doctor.

**C Walsh** stated that she and her mom had to do their own research and it was really hard to find one. She added that finding a doctor to take you is one thing, but finding one that is the right fit isn’t always easy.

**T Pasquini** asked if she could explain some of the differences she experience going from pediatric care to adult care.

**C Walsh** stated that in pediatric care the doctor and team spend more time with you. They get to know you better.

**R Thompson** – asked about advocacy. He stated that it sounded like your mother was a great advocate for you. Could you comment about that and could you think of how we could help those that don’t have an advocate like your mom.

**C Walsh –** She stated that her care is very complex and she couldn’t imagine how hard it would be to manage her care and all her appointments without a point person like her mom. She would recommend that a case manager assigned to a rare disease patient would be very helpful. When care teams have a case manager, it makes things a lot better for both the patient and the provider.

**D Tierney –** Thanked Caroline for sharing her story and invited her to stay and participate in the discussion if possible.

He then asked Representative Joe McKenna if he would like to introduce Congressman McGovern.

**Representative McKenna** introduce Massachusetts Congressman James McGovern, who sponsored a bill on Medical Nutrition. He introduced the Medical Nutrition Equity Act HR 6892 last year and he will give us an update on the bill and how it may impact Massachusetts residents with a rare disease. He added that the bill has bi-partisan support and a number of other states have passed similar bills.

**Congressman McGovern** thanked the council for its work and for inviting him to speak. He spent the next few minutes giving a background on the bill and emphasized that the cost of medically necessary nutrition products can cost as much as 4 or 5 times more than regular food. He added that this bill would require Medicare, Medicaid, and the Children’s Health Insurance Program (CHIP) to pay for medical nutrition. He stated that the bill started out requiring all insurances, including private to be required but the logistics and committee participation would slow it down so they decided to drop the private insurance piece.

He stated that everyone with a digestive or metabolic disorder who needs medical nutrition should be able to have access to it.

He gave a link to the bill and stated he was open to any comments or recommendation on the bill language. He encouraged anyone to reach out it his office if they had comments. He then asked if anyone had any questions.

He then offered to partner with the Massachusetts RDAC to increase awareness about the issue.

**Rep. Livingstone –** thanked congressman McGovern for his work on this issue. He then asked if there was anything we could do in Massachusetts to support his work.

**Congressman McGovern-** stated that he believes that food is medicine and if there is a specific food that someone needs for survival, they should have access to it. He stated that he would be interested in partnering with the RDAC if there was ever an event on the issue.

**R Thompson –** stated that sometimes the paperwork that a provider is charged with completing is overwhelming. He asked if a provider stated that a specific food was medically necessary would it be accepted or would the provider still have to complete mountains of paperwork to get it approved.

**Congressman McGovern** stated that one of the goals of the bill was to streamline the approval process for providers. He added that he never understood why nutrition wasn’t part of healthcare. If someone needs specialized foods to live they should be covered.

**G Ryan –** as a payer, it would help if the bill specifically identified what should be covered.

**S Cook –** stated that she was a provider at Boston Children’s Hospital Complex Care unit and she wanted to note that thickeners are usually not covered by insurance and they are very expensive. Many children need the thickeners in order to swallow liquids.

**Congressman McGovern –** Thanked the RDAC for the invitation to speak and stated that anyone could reach out to his staff person, Bella if they had any comments or suggestions about the bills language. Bella’s email is Isabella.eddo@mail.house.gov

**D Tierney –** then introduced Andrew Dwyer, PhD, FNP-BC to speak on the Challenges for a Rare Disease Patient Transitioning from Pediatric Care to Adult Care**.** He let all know that Andrew was an Associate Professor of Nursing at the William F. Connell School of Nursing at Boston College who has dedicated his career to caring for individuals and families affected by rare diseases, researching the genetics of rare endocrine conditions, and co-creating solutions to help improve patient health and quality of life. Andrew will provide some background on his work in transitions of care from pediatrics to adult care and then facilitate a discussion the topic.

Andrew presented the following slides:

**Outline**

* What is transition?
* Barriers to smooth transition
* Examples of transition navigation
* Take home points
* Discussion

**What is transition?**

* Purposeful, planned process beginning at age 16
* Transition from pediatric care that is child/family focused care that involves dependence to adult care that requires autonomy

**It is a process NOT an event**

A Dwyer then review his work stated core elements of a successful transition

* Age 12-14 – transition discussion begins
* Age 14-18 – need to track progress, assess readiness and develop a plan
* Age 18-21 – Begin transfer of care from pediatrics to adult care
* Age 18-23 – Complete transfer

**Key recommendations**

1. Develop a comprehensive structured program
2. Begin discussions with patient and family early
3. Foster communication between pediatric and adult providers
4. Implement a toolkit and tracking system to monitor progress
5. Dedicated staff for transfer (coordinator)
6. Involve the family. Use knowledge to empower patient and family
7. Use targeted therapeutic education to assist in transition success

A Dwyer then discussed a study in Europe and shared some lessons learned.

He stated that navigation was successful when the above recommendations were followed. Patients developed independence and adapted to their new environment, they learned the skills in decision making and self-management through targeted education sessions, and they were able to learn new coping skills to acclimate to a more independent adult life.

**Take Aways**

* Process vs. single event
* Continuity of care is important
* There are many complicating factors
* (patient, family, providers, health system)
* Structured, planned transition can help improve outcomes
* Collaboration & communication are key
* Resources are available to help you prepare
* Technology (m-health) offers new opportunities to support
* transition

**A Dwyer** then asked if anyone had questions.

**T Pasquini** asked if families had any trouble finding an adult provider to take them?

**A Dwyer** suggested that it may be helpful to have a list of adult providers willing to take on complex rare disease patients. He suggested that we may want to work with advocacy groups to find providers for specific conditions.

**N Grant** added that he thought that having a list of providers would be helpful. He also added that another issue that he has found with his brother’s care is the lack of small sized equipment in adult care environments. He stated that many rare disease patients are of small stature and in adult environments, they don’t usually have pediatric sized equipment. He added that we couldn’t forget about the structural gaps that exist in our healthcare system, like the lack of pediatric equipment.

**A Dwyer** stated that ergonomics situations were definitely a concern. We will need to brainstorm about possible solutions.

**G Thomas –** stated that she has heard from families that once their child goes to adult care they no longer had access to their medical records.

**A Dwyer –** stated that orphan drugs or the repurposing of drugs in pediatric populations may also be an issue. Pediatric clinical trials are very difficult.

**Rep McKenna** asked what the practical outcomes were from his work.

**A Dwyer –** stated that the implementation framework was the most practical outcome. He also emphasized that most likely the biggest problem is engaging adult providers. He recommended engaging Department Heads in the process. He also noted that the US healthcare system may have too many structural barriers to overcome.

**D Tierney –** thanked Andrew for his presentation and stated that although this was a wonderful model, it was a research study and how could we learn from this and think about recommendations for the RDAC.

**A Dwyer –** stated that it may take some sort of partnership in the US healthcare system.

**T Pasquini –** asked if there was any movement in US medical schools to educate medical students about this type of framework?

**A Dwyer -**stated that in his work, he has noticed that rare disease patients and families become experts in their condition. Maybe we look to them to partner with other patients who have similar conditions.

**S Cook –** asked if the council could put institutional pressure on providers. If there was a way that a provider could not say no to accepting a rare disease patient. Could we make a recommendation that a provider cannot decline care to a rare disease patient?

**D Tierney –** asked if people may be interested in recommending a rare disease patient bill of rights?

**A Dwyer –** announced that Boston College holds a rare diseases symposium every year with a goal of reaching all departments. He added that community based programs are really important for rare disease patients. They should have to drive long distances to access services. They should be in their community.

**R Thompson –** stated that he would be interested in looking at other models. It is an important topic and is a complicated issues. He also noted that he felt that the biggest challenge is payer based.

**S Cook** added that access to adult care is a huge problem. If there is a way to help providers work together it may help bridge the gaps.

**A Dwyer** – asked if it would be possible to convene a group of thought leaders to brainstorm and come up with some recommendations.

**C Achkar –** stated that she felt strongly that the discussions about transitioning to adult care need to start early and with the pediatricians. She added that many rare diseases are unique and it’s not realistic to think that adult providers can be an expert in every rare disease. Most patients and families become experts in their condition. She also felt that with support, any adult provider should be able to manage rare disease patient care. Most rare disease patients should be able to be cared for in their community.

**D Tierney** stated that we were at our time limit but we will continue this discussion. He thanked Andrew for a very productive presentation and discussion.

He then asked for the subcommittee chairs to share an update with the council.

**G Thomas (Policy Subcommittee co-chair)** stated that the policy committee will be meeting quarterly. They have had one meeting where they established their goals and objectives for the year. Their next meeting was on April 9th.

**T Pasquini (Research Subcommittee chair)** reported that the research subcommittee would be holding their first meeting this month. They will be discussing the burden of rare disease study and making suggestions on how to move forward.

**D Tierney (Steering Committee chair)** stated that the committee discussed the information line, ombudsman topic and office of rare disease, however did not come up with any recommendations at this time. He then added that most of the last meeting was related to council engagement. He introduced Tai to speak to the topic.

**T Pasquini –** stated that all of the council members have very busy schedules. All have expertise in specific areas and it may be helpful to survey the council members to see what kind of time they have to commit and what their interests are in their council work. She stated that she worked with Dylan to come up with a short survey and they would be sending it out shortly.

She hoped that all would take a few minutes to complete the survey.

She then added that she would be attending the World Orphan Drug Conference in Boston and would be interested in meeting up with any council members that would also be attending.

**D Tierney** stated that we were approaching 11 am but he wanted all to know that the RDAC trust account was set up and within the next 2-3 months, we should be able to start accepting donations. He then asked for a motion to adjourn.

**Rep Livingstone** made motion to adjourn. **T Pasquini** seconded.

**D Tierney** asked if all were in favor of adjourning, all nodded. He asked if anyone was opposed. No one answered.

He adjourned the meeting at 10:59 am.

**NEXT FULL COUNCIL MEETING: Scheduled for May 22, 2025.**