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

 **RESEARCH SUBCOMMITTEE**

REMOTE MEETING: Wednesday March 19, 2025

**Meeting Time 9:30 am – 10:30 pm**

**Meeting Minutes –** approvedJune 18, 2025

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**Tai Pasquini, subcommittee chair,** welcomed all to the meeting. She let all know that this was the first meeting of the Research Subcommittee.

She then conducted a roll call to establish a quorum.

**Roll Call**

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|  | Member | Present |
| 1 | Dr Olaf Bodamer | X |
| 2 | Dr. Heather Gray-Edwards | X |
| 3 | Yue Huang | X |
| 4 | Andrew Dwyer | X |
| 5 | Tai Pasquini | X |

A quorum was established and the meeting was called to order at 9:32.

She then asked all members if they could introduce themselves and talk briefly about why they want to be on this subcommittee.

Each member introduced themselves to the group.

**T Pasquini** then shared the goal statement and asked if anyone had any edits, revisions or suggestions. She read the following statement:

“The goal of the Research Subcommittee is to guide and support the research activities of the Massachusetts Rare Disease Advisory Council (RDAC). The subcommittee will be responsible for identifying and synthesizing existing research to provide evidence for RDAC initiatives as well as leading and supporting new research projects or collaborations that seek to provide data related to the goals outlined by the RDAC legislative mandate, such as topics related to the social and economic burden of disease, prevalence, and data collection methods.”

**Dr. Gray-Edwards** asked for further clarification of the statement. She asked for clarification on the word synthesize.

**T Pasquini** explained that some projects may require using existing data or collecting and analyzing new data.

**A Dwyer** suggested editing the language to say “the subcommittee will be responsible for identifying and synthesizing existing research to inform RDAC initiatives”

**T Pasquini** asked if we then add the language “additionally, the RDAC may support new research project or collaborations”

Should we leave off the topics.

All agreed.

She then read the final version of the goal statement

 “The goal of the Research Subcommittee is to guide and support the research activities of the Massachusetts Rare Disease Advisory Council (RDAC). The subcommittee will be responsible for identifying and synthesizing existing research to inform RDAC initiatives. Additionally, the RDAC may lead or support new research projects or collaborations.”

She then asked if there was a motion to vote to accept this new language.

**A Dwyer** motioned to accept the revised language**. O Bodamer** seconded.

All were in favor, no one opposed.

**T Pasquini** then shared the some of the legislative mandates the this group would address:

* Dissemination of findings of the RDAC’s research activities
* Use of existing publicly available records and information to estimate prevalence and causes of RD receives and considers reports and testimony from experts and community-based organizations
* Develop methods to publicize the profile of social and economic burden of RD in the Commonwealth
* Determine the human impact and economic implications of early treatment of RD versus delayed or inappropriate treatment
* Evaluate the current system of RD treatment and develops recommendations to collect RD data (i.e., registry)
* Annually file a report with legislature including the current state of comprehensive RD plan, actions taken and progress made, and estimate the cost savings upon full implementation of the comprehensive RD

Next she asked the group to review the following objectives, acknowledging that the first one was completed:

* Establish sub-committee goals and meeting schedule
* Identify opportunities to collect data related to the pressing challenges facing the rare disease community and their caregivers in the Commonwealth
* Collect data to quantify and define the burden of rare diseases in the Commonwealth
* Provide recommendations for additional methods of data collection to further RDAC goals

She asked if the above bullets best describe our planned work.

**O Bodamer** asked if we were considering the undiagnosed patients?

His suggestion was to add the language “ *Identify opportunities to collect data related to the pressing challenges facing the rare disease community, including individuals who are undiagnosed, and their caregivers in the Commonwealth.”*

 **A Dwyer** asked if the group was considering reviewing existing data or collecting new data?

**T Pasquini** stated that we may use existing data or collect new data based on the project.

She then asked if the group could all agree on the revision and agree to the final version of the objectives.

1. Establish sub-committee goals and meeting schedule
2. Identify opportunities to collect data related to the pressing challenges facing the rare disease community, including individuals who are undiagnosed and their caregivers in the Commonwealth
3. Collect data to quantify and define the burden of rare diseases in the Commonwealth
4. Provide recommendations for additional methods of data collection to further RDAC goals

She then asked if there was a motion to accept the objectives as modified?

**A Dwyer** made a motion to accept the objectives as modified, **O Bodamer** seconded.

All were in favor, no one opposed.

**T Pasquini** then reviewed the mandate in the legislative language for defining the burden of disease in the Commonwealth.

Then, she reviewed the following rationale bullets;

* Identify the true financial burden on the family for direct, indirect, and non-medical and uncovered healthcare costs
* Aid in policies that can impact healthcare allocation decisions
* Generate solutions to decrease the burden on the family including identifying and connecting individuals to current programs
	+ This could include changing eligibility criteria or expanding program parameters
	+ Increasing education and finding new ways of reaching potential eligible participants
* Launching new programs or support services
* Fixing the healthcare system for our most vulnerable and challenging cases can help make improvements for us all!

RDAC chair, Dylan Tierney joined the call at 10:02

She then described the work to date on planning for a burden of disease stud and reviewed the below tables.

She reviewed the table and the discussion summary from the last full council meeting. She reviewed thoughts and comments from the council and then asked for this subcommittee’s comments and thoughts on how to move forward.

**H Gray-Edwards** stated that she believes that there would not be any interest in funding this type of study.

**T Pasquini** added that she felt that there may be interest in funding something like this. She added that there is no plan to move forward with Optum and stated that it was just a starting point. She then asked for discussion on the topic.



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**Y Huang** added that we should fully investigate the vendor to ensure that they have extensive experience using claims data. She stated that using claims data is very messy and time-consuming. She would want us to fully vet any vendor we chose.

She added that she recently had a meeting with MassBio and rare disease drugs were mentioned. Although there is a lot of cuts in federal funding, it seemed that they would be interested in supporting this type of study.

**H Gray-Edwards –** stated that we needed to include Medicaid data in any work we do.

**O Bodamer** agreed that we needed to include Medicaid data in any work, specifically because of the diversity necessary for gathering the full picture. He also noted that the study discussed did not include any ethnic or race data. He feels that is very important to include race and ethnicity data in our work to accurately reflect the full picture of the rare disease burden. He also noted that the study only includes approximately 400 diseases.

**H Gray-Edwards** asked if we could reach out to advocacy groups and ask if they would share their registry data.

**T Pasquini** stated that we could definitely could use data sources for other organizations. However, in her experience, this data often does not follow traditional and conforming data collection practices.

**D Tierney** added that this type of study could be a “one of a kind” study that other states could model. We should be clear about the benefits of this type of study? He also noted that if we decide to go outside to solicit this work, we would need to go out to bid. It would have to be a competitive bid process.

**T Pasquini** asked if all were in agreement to pursue this project.

**D Tierney** stated that this work was mandated in our legislation.

**H Gray-Edwards** stated that this was a very worthwhile and important project.

**A Dwyer** agreed that this was a very important project and agrees that we should do the most comprehensive project to give us the best information on the true burden of disease. He added that we needed to think broadly and include life-threatening and life-altering diseases. He feels strongly that we need to include qualitative data in the project.

**T Pasquini** asked if we should seek other bids or should we go with the one we have, Optum.

**Y Huang** stated that she believes that our decision may depend on the funding source. She asked what our strategy is on moving forward.

**T Pasquini** summarized by offering to write up a one-page document to describe what we want to do and send back to the group for comment.

She let all know that time was coming to an end, and the next meeting of the subcommittee is

 June 18th, 2025 from 9:30 AM – 10:30 AM

Then asked for a motion to adjourn.

**A Dwyer** motioned to adjourn

**Y Huang** seconded

**T Pasquini** adjourned the meeting at 10:31