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

**Workgroup 1 Subcommittee Meeting**

Monday, April 3rd, 2023

2:05 PM – 2:55 PM

**MEETING MINUTES**

**Approved June 5, 2023**

**Welcome**

**D. Miller** welcomed all to the meeting. The meeting was called to order at 2:05. D. Miller read a short statement asking all guests to put their cameras off and mute their microphones if they were not on the agenda or part of the committee.

He then proceeded with roll call.

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| **Subcommittee member** | **Present** |
| **David Miller (Chair)** | **X** |
| **Ali Mahady** | **X** |
| **Andrew Dwyer** | **X** |
| **Andrew Lane** | **X** |
| **Tai Pasquini** | **X** |
| **Ann Wessel** | **X** |
| **Celia Segel** | **X** |
| **Julie Gortze** | **X** |
| **Ryan Thompson** | **-** |
| **Jeff Livingstone** | **X** |

**D Miller** asked if everyone received the minutes from the last meeting and if anyone had any corrections. No one had any corrections. He then asked if there was a motion to accept the minutes as written.

**T Pasquini** made a motion to vote to accept the minutes as written

**A Dwyer** made a second

**D Miller** conducted a vote to accept the minutes

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| **Subcommittee member** | **Accept** |
| **David Miller (Chair)** | **X** |
| **Ali Mahady** | **X** |
| **Andrew Dwyer** | **X** |
| **Andrew Lane** | **X** |
| **Tai Pasquini** | **X** |
| **Ann Wessel** | **X** |
| **Celia Segel** | **X** |
| **Julie Gortze** | **X** |
| **Ryan Thompson** | **-** |
| **Jeff Livingstone** | **X** |

**D Miller** opened the meeting by summarizing the work of the committee to date. He thanked everyone for the work.

**T Pasquini** shared that she sent the google doc with all of the summary work from people to the full committee for review.

**D Miller** shared the outline with the group and asked if anyone had any thought on the introduction.

**J Livingstone** asked if anyone knew of any other rare disease initiatives that were happening in Massachusetts. He expressed that knowing the history of rare disease efforts in the state could help support our initiative. He thought that it was important to state in our introduction, why rare disease prevalence was important. He shared that including a little bit about rare disease history and why the RDAC was formed is important. Should that be in the introduction? We should at least let the reader know that it is very difficult to get rare disease data.

**D Miller** asked the group if anyone knew where to find this kind of information

**J Livingstone** asked if it was possible to ask the legislators on the RDAC if they know how the legislation was created.

**T Pasquini** added that the only collaborative work she knew of was the workaround Rare Disease Day. She did not know of any other rare disease initiatives to determine prevalence. She did state that she thinks other states may be trying to do the same thing. She also stated that some rare disease organizations have determined the prevalence of their specific disease or disorder but she does not recall anyone looking at rare disease prevalence for all rare diseases.

**J Livingstone** suggested that maybe it would be important to state that in our introduction.

**D Miller** asked if all could look at the next section of the outline. This is mostly referencing definitions and the importance of this work. Section 2b addresses the importance and 2c addresses why international prevalence numbers are important and helpful for our work. We want to explain here why we are using this data to help us validate our own research data. We want to let the reader know the rationale that we use to measure MA data to global estimates. He asked the group how we could show the comparisons in the paper.

**T Pasquini** noted that in some of the other data uses ranges. She asked if we wanted to use ranges to address the differences. She also noted that she has been working in rare disease science for several years and the interest in determining rare disease prevalence is a fairly new interest.

**D Miller** noted that the audience for this paper is the state legislature. He asked the group if it made sense to seek some feedback from the legislature before we finalized the paper. For example, sending them a draft before it’s made final.

**A Lane** added his thoughts. He thinks that the outline was good. We should start with the introduction to identify the importance of defining prevalence.

**T Pasquini** added that she was not confident that some of the US numbers were not accurate so we need to validate with others. Some of the European numbers seem more validated.

**A Lane stated** that he ran into similar issues finding US data. He called the FDA and was told that they don’t keep any prevalence data. They rely on the grantees to submit the data. He then asked if the paper was based on our validation and the comparative range we use to make the determination.

**T Pasquini** stated that some the diseases that we want to include do not have good data. She asked if people thought we should reach out to Orphanet? She also stated that she would reach out to NORD to see if they know of any other states that are doing what we are.

**D Miller** asked if it would be sufficient to just state in our paper that we think the Orphanet data is the most robust and that is why we used it to calculate MA prevalence.

**A Lane** stated that we already used some of the Orphanet data to do some of our calculations.

**T Pasquini** stated that she thought that we should extrapolate using the Orphanet data

**J Livingstone** stated that he thought that one point we hadn’t discussed was personalized medicine. The rare disease world is really looking at personalized medicine. How do we address that in our paper?

**A Mahady** asked how we account for all the people that come to MA for treatment of rare diseases and the many people that are still undiagnosed.

**D Miller** stated that he thought the best way to address that was by citing publications that state the percentage of people that go undiagnosed every year. There is some pretty good data on that.

**K Mills** added that maybe we should address the caveats and limitations of our work at the beginning. Letting the reader know up front that the numbers may be lower than determined.

**T Pasquini** asked if we should include the burden of disease. I know we discussed this and it could muddy the waters so what do people think about it.

**D Miller** asked if we should just write a comparison piece that addresses the burden. We could finish this prevalence work as we plan and just touch on the burden. After we complete this work we could write a companion piece about the burden of rare diseases. I think it may be too much to take on both at this point.

D Miller then asked who would be willing to help write a section. Tai has done a lot of the work to date so is there someone else who can help?

**A Mahady** added that she did not have the background to write a section but she was willing to help in other ways. She would be willing to read and edit pieces.

**J Gortze** added that she felt the same. She wasn’t qualified to write sections but was willing to help in other ways. She also wanted to make sure that we had some way of capturing all the people that come to MA for rare disease care.

**J Livingstone** offered to help. Just let me know how you want me to help

**D Miller** asked all to send any thoughts or comments to him by Monday, April 10th. He wants to send a draft to legislators within 2 weeks.

**A Lane** walked through the basics of using google docs so that people could feel comfortable sending information directly to the documents. He assured everyone that they couldn’t delete or mess anything up. Just add what you can.

**T Pasquini** added that she was happy to work on the definitions and challenges section. I can work on the determination of numbers but I would need help with that.

**D Miller** added that he would work on the numbers determination piece.

He then stated that we were at the end of the meeting. He asked for a motion to adjourn.

**A Lane** made a motion to adjourn

**A Wessel** made a second

**D Miller** adjourned the meeting at 2:58 pm