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

**Workgroup 1 Subcommittee Meeting**

Wednesday, February 8th, 2023

3:00 PM – 4:00 PM

**MEETING MINUTES**

**Welcome**

**D. Miller** welcomed all to the meeting. The meeting was called to order at 3:02. D. Miller read a short statement asking all guests to put their cameras off and mute their microphones if they were not part of the committee.

He then proceeded with roll call.

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

**D. Miller** asked if each person could give a brief introduction and what they would like to contribute to this workgroup.

**D. Miller** started by letting all know that he is a clinical geneticist with a lot of experience working with rare disorders.

**A Dwyer** let everyone know that he is a nurse practitioner and clinical investigator for rare disease research, mostly focused on endocrinology

**A. Wessel** introduced herself as a dietician specializing in metabolic disorders that are treated with diet

**T Pasquini** introduced herself as PhD in public health. She works as the Research and Policy Director for Congenital Hyperinsulinism International (CHI), where she helps facilitate drug access programs.

**A. Lane** introduced himself as a physician-scientist working on blood cancers. He stated that he mostly works in the lab but has done work on clinical trials. He also stated that all blood cancers are considered a rare disease.

**C. Segel** stated that she is the Associate Director of the Health Policy Commission Pharmacology pricing and policy group. She mostly works on the cost of drugs and how well they work. She monitors spending trends in pharma spending in Massachusetts.

**D Miller** thanked everyone and stated that he wanted to ground everyone to think about the main objectives of this workgroup. He put the legislative mandates on the screen for all to review. (below)

These are the specific charges that our workgroup is supposed to work on.

* “using existing publicly available records and information, undertake a statistical and qualitative examination of the prevalence and causes of rare disease to develop a profile of the social and economic burden of rare disease in the commonwealth”
* “research and determine the most appropriate method for the commonwealth to collect rare disease data, including a database for all rare diseases identified in the commonwealth along with known best practices for care of said diseases and such additional information concerning these cases as the advisory committee deems necessary and appropriate to conduct thorough and complete epidemiological surveys of rare diseases subject to all applicable privacy laws and protections”

He asked if everyone had a chance to read it. All agreed. He then stated that one of our main charges is to identify the prevalence of rare disease in MA.

How do we come up with this number?

**T Pasquini** – My thoughts are that maybe we produce a white paper. I also think that we need to consider burden. I think we also need to look at the impact on families and caregivers.

**D. Miller** – asked Should we produce one white paper with 2 sections or 2 separate white papers? One on prevalence and one on the burden of disease, both socially and economically.

**A. Lane** – Finding prevalence sounds simple but we know it’s not. There is something to be said for keeping it simple, though. Maybe we start with prevalence and then look at the impact.

**D. Miller** – Maybe it doesn’t matter if it’s one paper or 2. Let’s focus on the work right now.

**C. Segel** – I think it would be nice to have 3 sections;

1. What is the prevalence of rare disease in Massachusetts?
2. What it the impact on the quality of life for those with a rare disease?
3. What is the impact on caregivers of those with a rare disease?

I also wanted to let the group know that I have access to claims data. It’s summary data but it could be helpful. I can ask permission to access the summary data we need.

**D Miller** – I think the first thing we need to do is develop an outline for these white papers. One for prevalence and one for impact.

**C. Segel** – If we have ICD 9 or 10 codes, I can inquire about the data. The problem I think will be that there is not an ICD code for all diseases.

**D. Miller** – Would it make sense to select the codes or conditions that are among the most common. We could look at those to start.

**T Pasquini** – A number of studies look at the prevalence of specific rare diseases. We could look at those studies and extrapolate. Even the statistic of one in 10 is hard to trace back.

**C. Segel** – We also need to remember that there is a data lag. The latest full year of data is from 2020. Maybe we should use 2019 data to be safe. The only challenge may be looking at diseases that have had new treatments.

**D Miller** – Does it make sense to identify the rare diseases that have good data on prevalence? If we find this data we can spot-check against our numbers.

**A Dwyer** - I think that the legislature is looking for the overall impact on Massachusetts residents. I don’t think the prevalence number has to be perfect, it just needs to reflect the best number for the legislature to understand how many people in MA are impacted by rare diseases. We want to show them that we have done our best to get the most realistic number.

**T Pasquini** – I did my dissertation in 2019 so I have a lot of literature and data on orphan drug spending, caregiver burden and prevalence. I’m happy to share this.

**D Miller** then asked **T Pasquini** if she would be willing to draft an outline for all to review.

**T Pasquini** agreed to send out a draft outline within the next week or so. She also stated that she would like a medical provider, patient and caregiver to give their viewpoint for the paper.

**D. Miller** stated that he could write an introductory paragraph to frame the issues of prevalence and impact.

**A Lane** suggested that we identify the dominant rare diseases, and then each of us take one to do research and see if we can find prevalence numbers for that disease.

**D Miller** asked how we would find a list of the most common rare diseases in MA.

**A Dwyer** stated that he just googled and found a list. Could this be a good list to start with?

**A Lane** said that each person could take one of the diseases on the list.

**D. Miller** asked for **A Dwyer** to read the list.

The list is as follows:

* Ehlers-Danlos Syndrome (EDS)
* Sickle cell anemia
* Cystic Fibrosis (CF)
* Hemophilia
* Duchenne Muscular Dystrophy (DMD)

The UK also includes Fabry Disease and spinal muscular atrophy (SMA)

**D. Miller** – I’d like to include SMA as it’s been in the news recently. I’m happy to look into DMD and SMA

**A Wessel** agreed to look into Hemophilia (Mary Lou will send Ann the number for Rich Pezzillo, who has hemophilia and is the executive director of the New England Hemophilia Association. (NEHA)

**A Dwyer** offered to look into EDS

**A Lane** offered to take Sickle Cell

**C Segel** offered to look into CF

**T Pasquini** stated that she also volunteers on the regional group for NORD and they have a list of the organizations who may have data on some of these diseases. People may want to check the NORD website for reference.

**D Miller** asked everyone to collect their research and send it to **T Pasquini.** She will organize the information and we can review it at our next meeting.

**D Miller** then asked if we could agree on a regular time to meet every other month. Maybe it makes sense to send out a doodle poll to find the best time.

Mary Lou agreed to send out a doodle poll to try to find the best time for us all to meet in April.

**D Miller** asked if **T. Pasquini** could report back to the full council at the March meeting as he will be out of town at a conference. **T Pasquini** agreed.

**D Miller** said that we were at the end of our scheduled time.

He asked for a motion to adjourn?

**C. Segel** a motion to adjourn

**T Pasquini** made a second

**D. Miller** asked if all agreed to adjourn. All agreed. The meeting adjourned at 3:59 pm.