

## Rare Disease Advisory Council

February 27, 2024 | 2:00-3:00pm

### Meeting Minutes

**Members present:** Lesley Bennett, Colleen Brunetti, Jim Carson, Mary Caruso, Kevin Felice, Joanna Gell, Emily Germain-Lee, Craig Miller, Saurabh Vaidya

**Members absent:** Dorian Long, Miriam Miller, James Rawlings, Michele Spencer-Manzon

#### Introduction

- Members introduced themselves and their role on the RDAC
- January 23<sup>rd</sup> meeting minutes were amended to reflect that James Rawlings was present and volunteered to be treasurer of the RDAC, amended minutes passed unanimously

#### Public Comment

- Guests introduced themselves and their connection to the rare disease community and advocacy
- Aida Carini shared her experiences living with a rare disease, especially the difficulties finding care for a disease that does not fit neatly into one specialty
  - She stressed the importance of medical treatment for individuals with rare diseases
- Adrienne Manning, director of CT's newborn screening program spoke about her work with the program
  - Diseases screened for by the newborn screening program:  
<https://portal.ct.gov/Newborn-Screening-Program/Pages/CT-NBS-Panel>

#### Update on Rare Disease Day 2024

- Leslie shared that the rare disease awareness event will be held at 9am on February 28, prior to the press conference with the public health chairs and the RDAC at 10am

#### Connecticut Rare Disease Forum, March 7, 2024

- JAX laboratories is hosting a rare disease forum next Thursday, March 7, from 2:00-4:30 p.m. with a reception beginning at 4:30 p.m.
  - Individuals involved in patient advocacy groups can register for free
  - <https://www.eventbrite.com/e/connecticut-rare-disease-forum-tickets-814172771677?aff=oddtcreator>
  - Reach out to [lisa.roy@jax.org](mailto:lisa.roy@jax.org) with any questions
- The event will have two panels on novel diagnostics and novel treatments, respectively
- Michele Spencer-Manzon and Craig Miller, RDAC members, will be on the two panels

#### Legislative Update--SB 175

- Sen. Anwar introduced SB 175, looking for funding for the RDAC
- Leslie gave an update on a bill that has been proposed in the Human Services Committee for rapid whole genome sequencing of infants, during the first year of life for critically ill infants
- Mary Caruso shared information on a bill related to access to diagnostic medical equipment (5200)
- Colleen raised the idea of having the RDAC advocate for having patient voices on relevant committees

### **Bylaw/Governance Overview**

- Leslie gave an update on the governance of the RDAC, and asked members for comments and feedback
  - She spoke with the chairs of the public health committee regarding funding for the RDAC, and granting the council the ability to solicit funds
    - Based on the current language, the group may only identify, not solicit funds
  - Leslie recapped legal suggestions that she received on the draft RDAC bylaws
    - One recommendation: three standing committees (such as Operations, Website and PR, Advocacy and Legislation), with ad hoc committees that come up
  - Proposed executive committee structure: two co-chairs, DPH representative, and two more (one health care provider representative, one patient representative)
- Colleen suggested clarifying that members can sit on multiple committees, and also suggested the creation of committee charters
- Colleen and Craig volunteered to help redraft the governance

### **In-Person Meeting Planning**

- Leslie asked the group for comments on holding a public hearing for rare disease patients
  - Colleen recommended having the patient portion before the RDAC's meeting, Emily agreed with this
- Leslie will speak with JAX offline to determine an in-person meeting in either March

### **Closing**

- Discussion of social media presence
  - Colleen raised questions about any guardrails from the state regarding the creation of an email list or social media presence – DPH is being consulted
  - Mary spoke about her organizing work and engagement via Facebook
  - Saurabh volunteered to help set up a Facebook page
- Colleen motioned to end the meeting; Saurabh seconded – the meeting adjourned at 3:00pm