

## Rare Disease Advisory Council

January 23, 2024 | 2:00-3:00pm

### Meeting Minutes

**Members present:** Colleen Brunetti, Craig Miller, Mary Caruso, Joanna Gell, Emily Germain-Lee, Michele Spencer-Manzon, Lesley Bennett, Dorian Long, Miriam Miller, Jim Carson, James Rawlings

**Members absent:** Saurabh Vaidya, James Rawlings, Kevin Felice (Lynn Ricci stood in for Dr. Felice at this meeting)

#### Introductions

- The members of the RDAC introduced themselves and their association

#### Public Comment

- Rare disease patients, parents, and other rare disease advocates introduced themselves as part of the public comment period
- Danielle, representing The Denise D'Ascenzo Foundation, shared information on a fundraising walk for rare diseases in April: <https://thedenisefoundation.com/walk-to-fight-rare-diseases>
  - Email Lesley for further information on this event
- Lisa Roy, representing the Jackson Laboratory (Jax), shared a save-the-date for March 7<sup>th</sup> for a CT rare disease forum

#### Purpose of Meeting

- Leslie summarized the November meeting topics; members voted to approve the minutes

#### Rare Disease Day 2024 Planning

- Wednesday February 28<sup>th</sup> from 8:30-11am in the second-floor atrium of the legislative office building (Address: 300 Capitol Ave #5100, Hartford, CT 06106)
- Lesley encouraged members who are interested in speaking at the event to send her an email

#### Discussion of Bylaws/Governance

- Lesley stressed the importance of having formal RDAC governance, and asked committee members for feedback/comments
  - Members thought the overall governance structure was good, but mentioned a couple items to finetune
  - She suggested forming a committee to iron out details in the draft bylaws and asked members to volunteer if they are interested
  - Members who volunteered: Colleen Brunetti, Jim Rawlings, Mary Caruso, Craig Miller, James Rawlings (volunteered to be treasurer)
- Executive committee

- Leslie proposed a five-member executive committee (two co-chairs, treasurer, secretary, and a state government representative)
- In this structure, each member of the executive committee will organize a subcommittee with other committee members and members of the public for additional support
  - Secretary committee (in charge of the website), Treasury committee (in charge of fundraising) Governance Legislation committee (co-chairs will be in charge of this)
- Lesley solicited feedback on holding a public hearing either an hour before or an hour after the RDAC meeting, so that members of the public can comment and discuss major issues
  - Michele suggested that these are held quarterly, and that members should have a sign-up sheet where only one representative per organization can speak
  - Members were generally supportive of looking into holding public hearings

### **Potential RDAC legislation for 2024 CGA session**

#### Rapid whole genome sequencing for certain newborns

- Leslie spoke about her advocacy for newborn screening and rapid whole genome sequencing
  - In the past, legislation for this was killed because it was mistakenly thought of as replacing newborn screening
  - Estimates from other states is that there are ~200 kids per year who can benefit from this
  - Even if there are not treatments being administered, Leslie talked about how kids are taken more seriously by physicians and can receive better care/have a better quality of life
- Emily Germain-Lee brought up some companies that will do free exome testing for an unidentified disorder under specific conditions, and although it is industry-sponsored, it could be something to do more research into

#### Other issues to examine

- Colleen brought up the impact that pharmacy benefits managers have on the high cost of medications

### **Closing**

- Craig Miller motioned to adjourn; Emily Germain-Lee seconded
- The meeting adjourned at 3:04pm