Rare Disease Advisory Council

June 24, 2025 | 2:00-3:00pm

Meeting Minutes

Members present: Lesley Bennett, Emily Germain-Lee, Joanna Gell, Adrienne Manning, Mary Caruso, Colleen Brunetti, Dorian Long, Jim Carson.

Members absent: Michele Spencer-Manzon, Craig Miller, Kevin Felice.

Introduction

- Lesley Bennett called the meeting to order at 2:04pm.
- Members of the RDAC introduced themselves and stated their role on the council.
- Lesley announced the passing of one of the RDAC members, James Rawlings.
- Mary Caruso motioned to approve the May 22 meeting minutes and Colleen Brunetti seconded. The minutes were updated to reflect the correct date (May 27 instead of April 22) and passed as amended.

Rare Disease Community Presentation: UConn Genetic Counselor Program

- Maria Gyure and Lila Aiyar, genetic counselors and leaders of the UConn Genetic Counseling Master's Degree Program spoke to the RDAC about their program.
- This new program aims to create a pipeline of genetic professionals.
- Maria spoke about how their program increases access to genetic counseling in two ways by hosting this program at a state institution in New England and also offering an asynchronous online modality so students can balance their responsibilities and needs.
- Maria spoke about how students receive a variety of clinical and non-clinical experiences during their master's.
- Lila Aiyar spoke about the field work rotation that is a part of the students' curriculum. In this enrichment rotation, students have worked with JAX, with Adrienne Manniing and the newborn screening team, and in patient advocacy. Students are not only learning the science, but learning about the importance of advocacy and the importance of this work, and where genetic counselors can fit in.
- Lila expressed that the program is always looking for more ways that genetic counselor students can partner with the rare disease community, so students can have exposure to the issues that are facing families and better understand how they can advocate for the community.
- RDAC members will connect UConn's Genetic Counseling Program with the organizations in the state, and patient support groups.
    - Mary Caruso also suggested connecting graduate students with her daughter, who runs a non-profit that intersects with rare disease advocacy.

Subcommittee Updates, Discuss Guidelines for Subcommittees

Awareness Committee

- Colleen Brunetti shared that the Awareness Committee is working on creating a map of rare disease resources in Connecticut, and how individuals can tap into these resources.
- There are around 100 patient groups, for-profits such as businesses, research companies, and biotech companies, non-profits such as hospitals, other research organizations, and academia. She expressed that the Committee is hoping for this map to be an educational tool for the legislature, but also to foster collaboration among these different groups. Colleen shared that a potential role for genetic counselors would be to help with reaching out to the different groups identified in the state, to build relationships among advocates and other interested parties.
- Six core people are working on the mapping project. The awareness committee is aiming to have the map project completed in time to write about in the RDAC's annual report.

Advocacy Committee

- Mary Caruso updated the group on the work of the Advocacy Committee.
- Four of the six RDAC legislative priorities passed
    - Funding for the RDAC – going to be working with DPH to work out the details of raising funds.
    - Expanding the Katie Beckett waiver program
    - Requiring insurance coverage for sickle cell gene therapy
    - Requiring insurance coverage for biomarker testing
- Lesley also mentioned the bill aiming to change the statutory definition of intellectual disability. The Human Services Committee agreed to not change the definition of intellectual disability without first consulting the rare community.
- The executive team will set up a date in July to discuss how to raise funds with DPH.
- Lesley spoke about emergency protocols and compassionate waivers: Currently there is a gap when children get off of the birth-to-three program but intellectual disability testing does not occur until around age seven, so a short-term conditional waiver for children in need is a priority of the RDAC.

RDAC In-Person Meeting Update: August, September, October

- August, September, and October RDAC meetings are scheduled for two hours, from 1-3pm.
- Craig Miller has arranged for the October meeting to occur at Boehringer Ingelheim in Ridgefield, CT.
- The September meeting falls over Rosh Hashanah. Lesley recommended moving the meeting back by a week to September 30th. Emily Germain-Lee is able to secure a room at UConn for that date.
    - Colleen motioned to move the September meeting and Mary seconded. The meeting will be moved from September 23rd to September 30th.
- Kevin Felice will check with Hospital for Special Care about a location for the August 26th meeting.
- Melia will extend the in-person meetings to be two hours. The September meeting may be from 1:30-3:30.

Member Terms: Members need to check with their appointing authority

- Lesley will reach out to legislators about the terms of each of the RDAC members. In each letter, Lesley will recommend that the first person appointed by each appointing authority serve a three-year term, and the second person appointed will serve a two-year term.
- Lesley may ask members to follow up with their appointing authority.

Discuss Possible Topics for Next Meeting

- The National Organization for Rare Disorders (NORD) has a meeting for RDACs in Washington DC from October 19th -21st. The Connecticut RDAC has two slots for members to attend. Members who are interested in attending the conference should reach out to Lesley directly.
- Global Genes is having a meeting on September 3rd in Boston; again, members should reach out to Lesley directly if they would like to attend.
- The group discussed what to do to memorialize the passing of James Rawlings, potentially putting together a donation to Michelle's House, supporting the sickle cell community.

Closing

- Emily motioned to adjourn the meeting and Colleen seconded. The meeting ended at 1:56pm.