Rare Disease Advisory Council

September 24 | 2:00-3:00pm

Meeting Minutes

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

Members absent: Kevin Felice, Craig Miller, James Rawlings, Michele Spencer-Manzon, Saurabh Vaidya

Introduction

- Lesley Bennett called the meeting to order at 2:03 pm.
- Members introduced themselves and stated their role on the RDAC.
- Lesley shared the purpose of the meeting:
    - Raising awareness of a number of medical conditions, several of which are rare diseases, as well as newborn screening.
    - Legislative and policy changes.
    - Annual report – due at the end of the month.
- Lesley summarized the August meeting and asked for any discussion or questions
    - Colleen Brunetti motioned to approve the minutes, and Emily Germain Lee seconded
    - The minutes passed without discussion

Public Comment

- For newborn screening awareness month, participants shared about the work Connecticut does.
    - Adrienne Manning shared that the DPH laboratory's newborn screening program has been in operation since 1964 where they screened for a single disease; now the lab can detect over 70 different disorders.
    - Karen Rubin added to Adrienne Manning's comment, speaking about the formation of the Connecticut newborn screening network, in communication with YNNH and DPH.
        - This network developed a fully electronic system to modernize the processes involved in newborn screening.
        - With these models and the collaboration, Connecticut newborn screening is at the forefront in the country.
- Emily Germain Lee shared about the conditions that she sees and treats in her role.
    - She has a clinic that follows the largest population of patients with Albright hereditary osteodystrophy – Connecticut is the focus of where these conditions are seen.

DPH Sickle Cell Disease Presentation and Q&A

- Alessandra Bogacki and her team presented on sickle cell disease (SCD), and the work that the Department does to identify, track, and follow-up with families.

- The sickle cell program encompasses both laboratory testing through newborn screening, and the short-term follow-up. DPH also spoke about identifying sickle cell trait, individuals that do not have SCD, but they are potential carriers, and they may experience health risks. Lastly
- Joanna Gell asked about the NBS efforts to reach out to healthcare providers in addition to families.
    - Marie Burlette shared that they are working closely with the Sickle Cell Disease Consortium to make these connections.
- Colleen shared a concern about adults with sickle cell disease being treated as drug seekers when they present in the ER in a pain crisis and asked if there was action done on equity and access to care around this.
    - Marie Burlette shared that there has been work done in the past outside of DPH to establish standard protocols used by emergency rooms when they are seeing individuals in crisis.

Legislative Issues/Update

- Lesley brought up PA 23-137, Section 4, which directs the Office of Policy and Management to consult with state agencies and councils to develop and recommend new state statutory definitions for intellectual disability and developmental disabilities. Because the RDAC was not yet established by the passage of this law, RDAC is not included in the conversation on these new state definitions.
    - Lesley spoke with OPM about this, since she believes the RDAC should have input in the report before it is published in January 2025.
- Mary Caruso shared her experience trying to get appropriate services for her children who have Friedrich's ataxia, expressing that she believes there is a gap in misunderstanding of the disease itself, especially since it is a rare and progressive illness.
    - She spoke about the universal assessment form used by DDS, DSS, and DoA for service, and highlighted aspects that were inaccessible, and potentially excluding individuals who need treatment.
    - She would like to see more flexibility in the criteria, a more wholistic care approach. One way to do this could be a layered evaluation process, where someone who fills out the universal form could indicate that it is a younger person, whose condition may progress overtime, that way younger patients are not as sidelined.
    - She would also like to see more education for the agencies on rare diseases.

Annual Report Discussion/Recommendations

- Lesley asked group members for recommendations on inclusions in the group's annual report, based on the outline she disseminated
- The group wanted to recommend forms be more inclusive towards progressive disorders

Closing

- Lesley shared several upcoming events in October related to rare disease advocacy
- Emily motioned to close the meeting; Mary seconded.
    - The meeting adjourned at 3:07pm.