

**Rare Disease Advisory Council**  
**Public Hearing and Council Meeting**

**May 28, 2024 | 1:00-3:00**

**Meeting Minutes**

**Members present:** Lesley Bennett, Colleen Brunetti, Jim Carson, Mary Caruso, Kevin Felice, Joanna Gel, Emily Germain-Lee, Dorian Long, Adrienne Manning, Craig Miller, Michele Spencer-Manzon

**Members absent:** James Rawlings, Saurabh Vaidya

**Introduction**

- Lesley opened the meeting at 1:05pm
- Members of the RDAC introduced themselves and their role on the RDAC to attendees

**Purpose of Meeting**

- Lesley stated that the purpose of the first half of this meeting is to hear from members of the public about issues facing the rare disease communities
- Lesley asked the RDAC to move the minutes to the council meeting portion of the agenda, all members present voted in agreement.

**Public Comment Session (1:00-2:30pm)**

- Two genetic counselors at JAX introduced their work at the laboratory and welcomed members to JAX
- Alison Leeds, Volunteer State Ambassador for Connecticut Rare Action Network
  - Kristen shared her story of being a caregiver to her husband with hereditary Angiodema with normal C1 inhibitor, and her 13-year-old son, with specific antibody deficiency. Alison focused on the complexity and challenges working to care for her son, namely, working with the school district to help her son access proper education services.
- Dean Houle, President and CEO of the MoyaMoya Foundation
  - David testified about MoyaMoya, a progressive rare disease, and his experiences with his daughter, who first had a stroke at age 19 during college. He emphasized the financial difficulties that patients undergo when they travel long distances to facilities for treatment.
- Fr. Nikolas Karloutsos, Assumption Greek Orthodox Church, Hospice Chaplin
  - Fr. Nikolas shared that his youngest child has Cardiofaciocutaneous (CFC) syndrome, and how this led to his entry into rare disease advocacy. He highlighted insurance reimbursement issues with his daughter's medical needs.
- Kristen Angell, Associate Director of Advocacy for the National Organizations for Rare Disorders (NORD), caregiver

- Kristen shared her story as a full-time caregiver for several family members, including her late father who died of pancreatic cancer, and her cousin Stacey who was diagnosed with cystic fibrosis (CF) at 18 months old. At the time, it was unheard of to hear of adults living with CF.
- She highlighted that individuals cannot get a double lung transplant in Connecticut – they have to travel to adjacent states. When her cousin was in New York, it was difficult both mentally and physically, because her core group of family members and doctors were not nearby and could not afford to travel every day or pay for the cost of a hospital-based hotel room.
- Kristen emphasized the importance of having access to information and resources of services for which people with rare diseases may qualify.
- Veronica Hernandez, caregiver
  - Veronica spoke about her experiences as a sole parent caring for her young daughter with lysosomal storage disease, neuronopathic Gaucher’s Disease Type 2 (nGD). She was told that her daughter would not live to age one -her daughter is currently 11 years old.
  - She spoke about the time required and the cost associated with getting care for her medically complex daughter, and how she has had to be resourceful to find the supports available to her. She received the Katie Beckett waiver after waiting for five years.
- Bryte Johnson, Government Relations Director for the American Cancer Society (ACS)
  - The ACS was the leading supporter of the biomarker testing legislation, and supporters of the rapid whole genome sequencing bill during the 2024 legislative session.
  - In the future, his organization hopes to expand biomarker testing coverage to private insurance
- Rep. Jillian Gilchrest, Connecticut State Representative
  - Rep. Gilchrest mentioned that a Katie Beckett working group was added to the rapid whole genome sequencing bill ([Public Act 24-130](#)). If signed, the legislature will populate the working group in August.
- Myrna Wantanabe, MS, PhD
  - Myrna spoke about her husband with sporadic inclusion body myositis (sIBM), and their difficulties finding care for him at home - she was able to get him into a Connecticut Veteran’s nursing home. She spoke about the mismanagement of his rare disease at a Farmington nursing home.
  - Myrna advocated for a file that accompanies a patient, labeling them as a patient of a rare disease and listing what that disease is, similarly to how individuals with allergies are labeled.
- Rachel O’Grady, MSW, Advocate at NORD and a Rare Patient
  - Rachel spoke about her rare conditions, diagnosed as an adult five years ago: Tethered Cord Syndrome and Spina Bifida Occulta. She highlighted her many issues fighting with her insurance, and she hopes the RDAC can make it so that private insurance companies do not deny rare patients

## **RDAC Meeting**

- Lesley ended the public comment and invited members of the RDAC to reintroduce themselves

### **Review/Approval of Minutes**

- Colleen motioned to approve the minutes and Craig seconded
  - The group passed the minutes unanimously

### **DSS/Medicaid Waiver Discussion with Christine Weston**

- Christine Weston, Director of Community Options at the Department of Social Services (DSS) gave a presentation on DSS home and community based services (HCBS)
- HCBS can be broken down into the state plan services, and the HCBS waiver services
- Christine emphasized that Medicaid must provide a comparable level of services and supports in the community as it is willing to cover in an institutional setting
- She also highlighted the state's portal for long-term care in Connecticut, called My Place CT
  - <http://www.myplacect.org/>
- The slideshow will be made available to RDAC members and posted to the DPH website.

### **Discussion of Bylaws**

- Craig, Colleen, and Lesley met a few times to edit the bylaws, and have distributed an advance copy to members
- Craig summarized the three sub-committees spelled out in the bylaws, and highlighted that they have designed the council to blend patient and healthcare representation
- The three encouraged members to review the document and leave comments by *June 15*

### **Conclusion**

- Lesley motioned to move the remaining agenda items to next month, all council members present agreed
- The meeting adjourned at 3:06pm