

Rare Disease Advisory Council
Public Hearing and Council Meeting

April 23, 2024 | 1:00-3:00pm

Meeting Minutes

Members present: Lesley Bennett, Colleen Brunetti, Jim Carson, Kevin Felice, Joanna Gell, Emily Germain Lee, Dorian Long, Adrienne Manning, Craig Miller, James Rawlings, Michele Spencer-Manzon

Members absent: Saurabh Vaidya, Mary Caruso

PUBLIC HEARING 1:00-2:00

- Rep. Mitch Bolinsky, 106th Assembly District of the CT General Assembly
 - welcomed everyone on behalf of the CGA, delivered opening remarks
- Julieta Bonvin Sallago, glycogen storage disease patient, Connecticut Children's
 - Spoke about personal experience with her rare condition, currently supports the glycogen storage disease program at CT Childrens
- Frank Tavarez-Mora, sickle cell disease patient, Quinnipiac medical school student
 - Spoke about his experiences living with sickle cell disease, including the stigma that comes with it
- Christopher Curren, MD advocate (www.kindnessovermd.org)
 - Spoke about experiences with his son who was diagnosed with DMD, as well as the need for early diagnosis/newborn screening both to help children with appropriate therapy and treatments, and to help parents understand more about the disease early on
- David Negron, caregiver of daughter with a rare disease
 - Spoke about his daughter with Tay-Sachs disease, and testified in support of SB 206 – expansion of the Katie Beckett waiver program
- Michael Negron, father of David Negron
 - Spoke about the need to reallocate money in the state budget to prioritize rare diseases
- Carolyn Macica, rare disease researcher at Connecticut Children's & Yale; coordinator Quinnipiac University Rare Disease events
 - Spoke about the transition from pediatric care to adult care for rare disease patients, and her work updating a transition care toolkit, and passed out fliers for an October rare event
- Yong-Hui Jiang, MD, PhD, director of genetics at Yale
 - Spoke about emerging technology for gene editing to help better understand rare diseases
- Elizabeth Olear, Senior Clinical Research Associate at the Yale Center for XLH in the division of Pediatric Endocrinology
 - Spoke about her rare disease research experience with XLH, and creating opportunities for patients to meet others with the same condition and learn more about their disease

- Megan Freeman, patient with 2q37 chromosome deletion syndrome
 - Spoke about her experiences living with a rare disease (around 170 people in the world have it)
- Phil Marella, father of two children with rare diseases, founder of Dana's Angel's Research Trust (DART)
 - Spoke about his experiences raising children with Niemann-Pick disease Type C
 - <https://danasangels.org/about/>
- Amy Chickles, ALS United Connecticut
 - Spoke about their organization's work supporting largely adult patients with ALS
- Jodi Gillian, president of BioCT
 - Testified in relation to SB8 - Applauds the intent of the bill (lower the cost of drugs for patients), but spoke about three components of the bill that they find troubling
 - 340b mandate – federal legislation – we do not know where the discounts are going, and often times they are not benefitting the patients
 - Prescription Drug Affordability Board (PDABs), – could make drugs expensive for pharmacies and hurt the community
 - Canadian importation – Canada is not very open to importing drugs, unsure how this would work
- Dominic Cotton, parent spouse and brother of many family members living with rare diseases
 - Spoke about his advocacy for rare diseases and gave his appreciation for the number of experts that come out to support the community and inform legislators
- Pamela Johnson, research auditor at Hartford Healthcare, rare disease patient and parent
 - Spoke about her passion and advocacy for rare disease and shared that she has hereditary angioedema

RDAC MEETING, 2:00-3:00pm

Introductions

- Members of the RDAC introduced themselves and their role on the RDAC
- The group approved the previous meeting minutes

Bylaws

- The bylaws subcommittee will get together in the coming weeks, more updates to come

Legislative Updates

- Lesley updated members on the four bills that members of the RDAC are watching: HB 5357, SB 206, SB 175, SB 306 – the first three bills are held up in negotiations
- Funding for the RDAC: Lesley brought up the creation of an independent non-profit to help finance the RDAC, will communicate with DPH to figure out a way to do this
 - If SB 175 does not pass, the RDAC cannot solicit funding
- The group went over the process for legislative session and the biannual budget
- Related to HB 5321: The Advisory Committee on Heritable Disorders in Newborns and Children may hold a vote to add DMD to the RUSP around May 9
- Lesley gave updates on additional bills:

- SB 1 contains a provision supporting Parkinson's disease
- SB 8 prescription drug affordability boards – a lot of these boards will hurt the rare community by denying access to certain necessary medications, because the boards do not have representation from individuals who prescribe medication to patients
- SB 307: Biomarker testing – passed the senate, moving to the house
- Healthcare access for people with disabilities: one room in clinics for people who use wheelchairs

Katie Beckett Waiver: Overview by DSS

- Paul Chase from the Department of Social Services gave an overview of the Katie Beckett waiver and answered questions that council members had. Key points included:
 - CT has ten different Medicaid waivers (1915C) that target specific populations, this is more than any other state
 - Katie Beckett = most unique waiver, allows participants to access Medicaid
 - It serves medically fragile community, ages birth-22, with no list of approved qualifying conditions
 - There are currently around 312 people on the waitlist for the program
 - There are a lot of people who are on the waiver who do not use the Medicaid services at all – they use it as a backup in case of a catastrophic event – these people should still be able to use these spots, but it's a difficult decision for members to decide to take the waiver
 - A majority of states operate Katie Beckett as a state plan option, instead of a waiver (Creates a coverage group for disabled individuals under the age of 18)

Closing

- The next meeting will be held at Jackson Laboratories
- The meeting adjourned at 3:02pm