Rare Disease Day 2025 at the LOB

Tuesday February 25 from 8:30am to 11am
2nd floor Atrium Legislative Office Building

Please join Senator Anwar and the Connecticut Rare Disease Advisory Council to Raise Awareness and Make a Difference in the Lives of Rare Disease Patients

AGENDA

8:30AM RDAC MEETING

- RDAC Member Introductions
- Legislative Priorities for 2025 session
  - Advocacy & Awareness Committee

9:00AM WELCOME & PRESS CONFERENCE

- Emcees—Dominic Cotton & Colleen Brunetti
- Senator Anwar, Rep. McCarthy Vahey & CGA members
  - Saurabh & Anay Vaidya-Hemophilia

9:15AM Speakers:

- Dr Rebecca Riba-Wolman—GSD (Connecticut Children's)
- Dr Pamrod Mistry—Gaucher Disease (Yale)
- Rachael Turner—Late Onset Tay Sachs
- Beth Frase (parent)—Pontocerebellar Hypoplasia
- Matthew O'Donnell—Alexion
- Shannon Belmont--GeneDx
- David Negron (parent)—Juvenile Tay Sachs
  - Kathy Flynn—NTSAD
- Dr Jessica Fennell—Pediatric Rheumatology (Connecticut Children's)
- Dr Uyen To—Wilson Disease (Yale)
- Bonnie Royster—Cornelia de Lange Syndrome (CdLS) Foundation
- Genetic Counselors: Elizabeth Charnysh & Alexandra Turhlar (Jackson Laboratories), Nivedita Rao (Yale)
- Dan Donovan—RareLife Solutions
- Courtney Coates—Hope In Focus
- Paul Pescatello—CIBA
- Sickle Cell speakers
- CLOSING--Pamela Johnson & Carl—HAE and NIH

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Rare Disease Info:

- In the US, a medical condition is considered RARE if it affects less than 200,000 individuals (ODA 1983)—in Europe a Rare Disorder is one that affects 1 in 2000
- According to NIH, more than 8, 000 Rare & Genetic Diseases have been identified affecting more than 30 million Americans—about 8-10% of the US population
- More than 75% of all Rare Diseases are due to genetic variations—about 20- 25% are due to infectious agents, toxins, environmental factors or acquired immune conditions
- NIH estimates that > 70% of those living with a Rare Disease are children—and ~30% of these children will not see their 5th birthday
- Except for asthma, obesity, and autism; all chronic diseases of childhood are considered rare—*including all pediatric cancers*
- Very few physicians are trained to recognize or treat Rare Diseases and there is very little reliable information or public resources for many Rare Diseases (Lack of awareness)
- It may take some patients >5 years to receive an accurate diagnosis since Rare Diseases are complex medical conditions, often presenting with symptoms of common diseases
- Less than 10% all Rare Diseases have a single FDA approved treatment
- Rare Diseases tend to be chronic, lifelong medical conditions that can be debilitating, life-limiting, slowly progressive or life-threatening—often requiring long-term care
- In 2019, NIH reported that the healthcare costs for Rare Diseases was approaching ~1 trillion dollars per year—costs that could be reduced with early diagnosis and treatment
- Cystic Fibrosis, Hemophilia, PKU, Tay Sachs, SMARD, Sickle Cell Anemia (US), Gaucher Disease, William's Syndrome, Niemann-Pick, Fragile X are all rare diseases
- In Connecticut more than 100 patient organizations, 50 companies, 3 medical schools, 4 hospital/academic centers, and Jackson Labs help those living with a rare disease
- NIH estimates that ~300,000 people living in Connecticut have a rare disease—*that is more than the populations of Stamford and Bridgeport combined*
- For 60 years the CT-Department of Public Health has been saving the lives of children born with certain (~70) treatable rare disorders through the *Newborn Screening Program*

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CT-RDAC Legislative Priorities for 2025

- Funding **(SB 562):** need ability to solicit funds for RDAC activities such as:
  - Creation of Website (resources) and Educational/Awareness materials
  - Survey of CT rare disease community
- Improving access to state waiver programs for children living with a rare disorder:
  - Expand the Katie Beckett Waiver--**SB: 801**
  - Create *Compassionate waiver* for critically ill children with rare disease--**SB 802**
  - Intellectual Disability Waivers: Children and families of children with rare disorders have a difficult time obtaining DDS waivers due to lack of awareness
    - Section 4 of PA 23-137 did not include RDAC or members or the rare disease community in any of the feedback group-**amend PA23-137-Sec 4**
    - Need to include RDAC in all discussions of a new definition if IDD, the need for IQ tests, and modernization of level of need tools used by DDS
- Emergency Protocols: legislation requiring emergency personnel to follow Rare Disease patient's specific protocol in an emergency situation or contact patient's physicians
  - Public Hearing on 2/26/2025 on **HB 6920**—please testify in emergent situation
- Ask DSS to submit application creating a Connecticut Sickle Cell Gene-Cell Therapy site
- Ensuring *access to Orphan Drugs* and all medications needed by Rare Disease patients
  - Amend **PA23-171**, Section 8 to include language protecting Orphan Drugs
  - Include language in new legislation providing some protections to Orphan Drugs used to treat 1 rare disease or up to 3 related rare diseases—**SB 566**

Patients speaking up to raise awareness of rare diseases and their effects on families, caregivers, employers, state programs, and our health systems is the KEY to making a difference!
My daughter Kelly was born with a rare metabolic disorder (classified as a metabolic epilepsy) and was not expected to survive her 3rd birthday because there were no cures or proven treatments for her unusual medical condition. Thanks to the help of number of wonderful physicians at Yale, Connecticut Children's Medical Center (UConn), Harvard, Johns Hopkins, and NIH we discovered an unusual dietary approach that may not have cured her disorder but allowed Kelly to control her seizures and enjoy life for almost 32 years! Please help all patients living with a Rare Disease and their families enjoy life by supporting research on rare diseases and development of new therapies for these conditions.

Thank you for attending our 2025 Rare Disease Day celebration at the LOB,
Lesley Bennett (CT-RDAC, Co-Chair)

- RDAC Members—Dr. Michele Spencer-Manzon, Dr. Emily Germain-Lee, Dr. Joanna Gell, Dr. Kevin Felice, Craig Miller, Colleen Brunetti, Mary Caruso, James Rawlings, Saurabh Vaidya, Adrienne Manning, Jim, Carson, Dorian Long, and L. Bennett