Rare Disease Advisory Council

October 22, 2024 | 1:00-3:00pm

Meeting Minutes

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

Members absent: Adrienne Manning, James Rawlings, Saurabh Vaidya.

DPH: Melia Allan.

Introduction

- Michele Spencer-Manzon called the meeting to order at 1:03pm and described the purpose of the meeting.
- Michele asked members of the RDAC to introduce themselves and state their role on the RDAC.

Public Comment

- The Yale Cystic Fibrosis Center presented on Cystic Fibrosis (CF).
    - Marie Egan gave an overview of CF, how the CF Care Team at Yale functions, and the research that the team conducts to study the disease and provide comprehensive quality care.
- Michael Weinstock presented on the Yale Hearing Loss program and pediatric otolaryngology, a rare disease that leads to hearing loss in children. He spoke about the work his care team does, and the potential for next steps, including genetic diagnosis and treatment.
- Dr. Daniel Glasser presented on Craniofacial Scleroderma at YSM/YNHH
    - This condition is a combination of inflammation and fibrosis in an autoimmune setting.
    - The center is involved in active collaborative research with three different centers around the country.
- Benjamin Tolchin spoke about functional neurological symptom disorder.
    - This is also labelled as Conversion Disorder in the DSM-5.
    - He spoke about the evidence-based treatments, including psychotherapy, physical and occupational therapy, and psychopharmacologic treatment of co-occurring psychiatric disorders.
    - The council asked if coverage is a barrier for these kinds of treatment – Dr. Tolchin confirmed that this is a barrier, and it is important to have coverage for these sorts of behavioral health treatments.
- Thomas Carpenter spoke on Metabolic Bone Disease in Children: X-Linked Hypophosphatemia (XLH).
    - This disease has a prevalence of 1/20,000, but it is the most common heritable cause of rickets. However, this is not only a childhood condition.

    - He shared some of the biggest problems and concerns: workforce issues – there are not many people trained to care for these patients; delays in insurance approvals for visits and medication; and necessity of coordinating complex multi-provider care.
    - He shared the members of the Yale Pediatric Metabolic Bone Clinic, the interdisciplinary care team for children with metabolic bone disorders.
- Laura Chen presented on Yale's Pediatric Aerodigestive Program.
    - Home nursing is a critical resource that these patients need.
- Sasha Zivkovik presented on the CMT Program and CMTA Center of Excellence at Yale.
    - Charcot-Marie Tooth disease is a hereditary motor and sensory disorder, affecting 1 in 2,500 adults. Over 100 genes have been identified as causal for CMT and related disorders, but most cases are caused by mutations in 4 genes.
    - In addition to genetic therapies, there are supportive treatments for CMT.
- Louise Lanoue spoke about the pediatric wing of the MDA care center.
- Yong-hui Jiang presented on Yale's Diagnostic Center of Excellence for the Undiagnosed Disease Network (UDN) Phase III, announced two months ago.
    - He highlighted that a significant fraction of rare diseases remains as undiagnosed diseases, but undiagnosed diseases may not be rare diseases.
    - The goal of phase 3 involves promoting a broader integrated and collaborative community across multiple clinical sites.
- John Kuster presented on primary immunodeficiency (PID) clinics at the Yale School of Medicine
    - PID is associated with clinically significant infections, sometimes associated with autoimmunity or inflammation.
    - He shared where the Immunology Clinics are located in the state.
    - Early diagnosis leads to cost-savings, but PID is under-recognized and PID diagnosis requires specialized testing. PID mechanisms are also not fully understood.
- Rima Fawaz presented on pediatric liver disease and the Pediatric Liver Transplant Program.
    - Their clinic has around 1500 visits a year – they have doubled their service in the last 2 years.
    - She shared information about their clinical research, trials, and funding.
- Cheyenne Beach spoke about the Yale Family Inherited Arrhythmia Clinic.
    - Cheyenne presented on the range of genetic arrhythmia syndromes, including those that are undiagnosed.
    - This clinic sees the family together in the same room (could mean 2 people, could mean 6 people!), to limit the amount of back-and-forth phone calls between relatives and give comprehensive care to the whole family at once.
    - Project ADAM is a national network of affiliates (mostly of children's hospitals) that work together to make schools as safe as possible. A lot of times this means creating emergency action plans, equipping schools with AEDs, etc. Yale is the Connecticut affiliate of Project ADAM.
- Jessica Sweeny spoke about the Cardiogenetics Clinic at Yale.
    - The clinic is looking to build out their referral network, specifically with orthopedics, ophthalmology, adult cardiologists, neurologists and psychologists, and GI.
- Katherine Kohari spoke about Maternal-Fetal Medicine – caring for people who are pregnant who have high-risk conditions, including rare diseases.

    - The Reproductive Genetic Counseling Program consult with pregnant people prior to diagnostic testing, interpret test results, and aids in insurance coverage.
    - The Fetal Care Center's goal is to identify patients who need transfer to the Yale New Haven Hospital for Children (YNHCH) vs those who can remain at the community hospital for delivery. They do various types of fetal therapy, including fetal gene therapy, and fetal enzymatic treatment.
- Vinita Knight presented on child neurology and epilepsy.
    - She shared some of the targeted treatments and therapies and highlighted the importance of newborn screening in their clinic.
- Alina Herlopian presented on the adult epilepsy clinic.
    - Patients who graduate from the pediatric clinic come to this clinic, in addition to adult-onset epilepsy.
- Jessica Srouji spoke about the Motility Clinic.
    - She described some of the associated conditions with motility and DGBIs.
- Monkol Lek spoke about the Neuromuscular translational program.
    - In addition to genetic research, his lab works closely with Connecticut-based patient foundations.
- Uyen To presented on the Wilson Disease Clinic.
    - Early diagnosis and treatment can help these patients lead normal lives. Patients experience delays in treatment as well as large expenses for genetic testing and treatments.
- Jon Koff spoke about the Adult Cystic Fibrosis program at Yale.
    - CF is an example of the success of centers of excellence. As a chronic disease, there is a huge need for mental health services.
    - PwCF have challenges with access and cost of medications.
- Jackie presented on the Yale Center for Bleeding and Clotting Disorders
    - They treat several conditions including hemophilia.
    - Resources needed include transportation, clinic space, genetic testing, access to clinics/blood draws, mental health support.
    - Other challenges include the access and cost of medication, and insurance issues.
    - In 2023, they saw 1800 patients.
- Kevin Hall spoke about pediatric cardiomyopathy, and the work that his team does to diagnose and treat patients in the state.
- John Stendahl spoke about the Yale Cardiomyopathy Program, which provides specialty care for inherited and infiltrative cardiomyopathies.
    - Hypertrophic cardiomyopathy is the most common condition treated, but the program treats numerous other conditions.

Closing

- For questions about the presenters/clinics/programs from today, contact Michele at michele.spencer-manzon@yale.edu to get connected with the correct individual.
- The meeting adjourned at 3:49pm.