Meeting Minutes

Joint Meeting of the
Rare Disease Advisory Council (RDAC)
Academic Research Institutions and Health Care Providers Subcommittees

Date: Tuesday, August 15, 2023

Time: 12:00 PM – 1:00 PM EST

Microsoft Teams Video Conference

Academic Research Institutions Subcommittee:

Member Name	Member Seat	Present	Absent
Dr. Barry Byrne	A representative from an academic research institution in Florida, which receives grant funding for research regarding rare diseases.	X	
Dr. Divya Patel	A physician who is licensed under Ch. 458 or 459, F.S., practices in this state, and has experience in treating rare diseases.	X	
Dr. Mustafa Tekin	A representative from an academic research institution in this state which receives grant funding for research regarding rare diseases.		X
Dr. Rajan Wadhawan	A physician who is licensed under chapter 458 or chapter 459 and practicing in this state with experience in treating rare diseases.	X	

Health Care Providers Subcommittee:

Member Name	Member Seat	Present	Absent
Dr. Anita Davis	A representative of organizations in this state which provide care or other support to individuals with rare diseases.		X
Rebekah Dorr	A representative of organizations in this state which provide care or other support to individuals with rare diseases.		X
Zana Dupee	An individual who is a caregiver of an individual with a rare disease.		X
Jessica O'Reilly	An individual who is 18 years of age or older who has a rare disease.		X
India Steinbaugh	An individual who is 18 years of age or older who have a rare disease.	X	

Guests: Kelly Rogers (Florida Department of Health), Tara Cockman (Florida Department of Health), Maggie Dilger (Florida Department of Health), Jon Conley (Florida Department of Health), Paula Libassi (Caregiver parent, Florida School for the Deaf and the Blind)

Call to Order, Member Roll Call – *Maggie Dilger, Florida Department of Health*
The meeting was called to order at 12:04 PM and roll was taken. A quorum was established for the Academic Research Institutions Subcommittee. A quorum was not established for the Health Care Providers Subcommittee.

Approval of Meeting Minutes – *Council Members*
Approval was not needed for the Academic Research Institutions Subcommittee's July 18, 2023, meeting summary. Because a quorum was not established, approval of the Health Care Providers Subcommittee's July 19, 2023, meeting minutes will move to the September 20, 2023, meeting.

Year 2 Objectives Implementation Plan
There was discussion of the Health Care Providers Subcommittee's recommendations with insight from the Academic Research Institutions Subcommittee.

Dr. Rajan Wadhawan discussed how NORD's Centers of Excellence designation focuses heavily on research and education and requires fellowship programs, clinical trials, etc. to obtain that classification. Dr. Wadhawan explained that there are many organizations in Florida that may have several elements to care for people with rare diseases but may not meet the rigorous requirements of NORD. He suggested there should be a subgroup of centers that may not focus on new discoveries and research but do focus on patients and families and clinical abilities. Within the council, there is a desire to create a "lighter" version of Centers of Excellence for Florida.

Discussion continued regarding the role of the council and its subcommittees. Dr. Wadhawan mentioned a document was shared that outlined NORD's scoring process for Centers of Excellence designation. Kelly Rogers with the Florida Department of Health mentioned the group needed clarification on who has authority for creating, developing, implementing, ongoing reviewing for the future (reapplying), and what specifically is the role of the RDAC for varying work that's being done in the state. Ms. Rogers mentioned there are other state councils that are being examined to see what their roles are and what processes have been established. The Department staff plans to research and conduct internal meetings and will report back to the council on their findings in the future. Ms. Rogers suggested this topic be discussed at the full council Meeting in October. The Department will reach out to Jon Hawayek to ask what other RDAC's are doing since he sits on other states' RDACs.

Regarding identifying and compiling screening tools, Dr. Wadhawan was unaware of a universal list and questioned if an all-inclusive list could be created since most conditions do not have defined treatment. Dr. Barry Byrne mentioned increased utilization of next generation sequencing as part of screening, but individual disease screening is not used as much because it is not economical. Rapid whole-genome sequencing (rWGS) is used but there are issues with cost. If screening is ordered during in-patient care, the cost can be covered under hospitalization costs. If screening is ordered in an out-patient setting, the cost can be a burden to the family. Obstacles can also come due to limited genetic counselors. It was asked if new technology for screening should be the focus instead of compiling a list of screening tools. Desk research will be conducted to see what other RDACs, agencies, and organizations are recommending as resources.

Dr. Byrne recommended there should be a single policy for advanced genetic testing/next generation sequencing through the Agency for Health Care Administration (AHCA) as this would simplify matters in the care of rare disease patients and shorten the diagnostic dilemma for patients. Dr. Wadahawan mentioned that even with new legislation, the issue of in-patient and out-patient coverage is a concern as well as a need for further definition from AHCA. It was decided to ask Medicaid leads to present on the new law regarding rWGS during the next full council meeting in October with the hope that a recommendation from the full council on this matter will occur.

While discussing surveying individuals with rare diseases to identify barriers in access to care for testing and treatment, it was mentioned that the biggest unmet need is where there is no defined treatment. Costs for whole genome sequencing can be an issue for some families though cost is decreasing. There is a burden in understanding the data received from sequence which can take a long time from genetic counselors or providers to identify variants. Organizations like Invitae will continue to data mine and update providers as variants are defined as being disease causing. The diagnostic rate is still very low, and there are still many unknown cases. These databases are used for researchers as well to see if any clinical findings can be compared worldwide.

Further discussion is needed to identify if there are opportunities to map out or if standardized processes already exist. The Department is requesting process maps from Newborn Screening as a resource.

Discussion moved to the Academic Research Institutions Subcommittee's recommendations. Dr. Wadhawan mentioned a list of geneticists in the state was created, but that list should be reviewed to ensure it was complete. The initial list compiled all board-certified geneticists, and the next step should involve reaching out to geneticists in the state to ask what their specialties are to be published on the state website (when created). The Department will come up with questions to ask those providers and send to Dr. Wadhawan for approval and feedback.

Kelly mentioned the Department is working with the communications team to begin the process of developing a website where all the recommended resources will be housed.

Public Comment – *Members of the Public*

Paula Libassi (Caregiver parent, Florida School for the Deaf and the Blind) thanked the subcommittee for its work. Kelly thanked her from joining the conversation and discussed the excitement of the RDAC and the meaningful work it's doing.

Adjournment – *Maggie Dilger, Florida Department of Health*

The meeting adjourned at 12:58 pm.

Meeting Reminders

The next RDAC Academic Research Institutions Subcommittee meeting is scheduled for Tuesday, September 19; 12:00 – 1:00 pm EST.

The next RDAC Health Care Provider Subcommittee meeting is scheduled for Wednesday, September 20; 12:00 – 1:00 pm EST.

The next Rare Disease Advisory Council (RDAC) Full Council meeting is scheduled for Wednesday, October 25; 12:00 – 1:00 pm EST.