Rare Diseases Advisory Council

April 12, 2024

Members in Attendance:

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	X
Department of Health and Environmental Control	Ginie Daguise (SCDHEC)	X
Department of Health and Human Services	Kevin Wessinger (SCDHHS)	X
Medical University of South Carolina	Dan Judge, MD (MUSC, cardiology)	
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	X
South Carolina Hospital Association	Kate Wink (Santee Cooper)	X
South Carolina Primary Healthcare Association	Vicki Young (SCPHCA)	X
Biopharma Industry	Jonathan Hawayek (SPARK Therapeutics)	X
Research and Treatment of Rare Diseases (3)	Maysen Mesaros, MS (MUSC, neuroscience) Neena Champaigne, MD (Pediatric Genetics) Chip Norris, PhD (MUSC, connective tissue)	
Patient (2)	Anthony Hamilton (familial TTR amyloid) Karen Kemper, PhD (scleroderma)	
Rare Disease Organization	Megan Onysko (CFF)	
Caregiver of person with rare disease	Jason Smith (hypoplastic left heart syndrome)	
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	X
Program Coordinator	Kendall Spillman (SCTR Success Center)	X
Guests In Attendance:		
MUSC - SCTR Success Center	Stephanie Gentilin (Director) Tara Pittman (Recruitment Manager)	X
Bleeding Disorders Association	Sue Martin (Executive Director)	X
Rare Disease Diversity Coalition	Taylor Dominguez (Project Manager)	X
Pharming Healthcare	Jessica Gordils (APDS Clinical Educator)	X
Acadia Pharmaceuticals	Kemi Olabisi (Rare Disease Pharmacist)	X
Artia Solutions	Sara Stolfus (Clinical Research Analyst)	X
NORD	Carolyn Sheridan (Policy Manager)	X
	Elizabeth Jones	X
	Tara Tsagouris	X
Alexion Pharmaceuticals	Bill Eicholzer	X

Meeting Minutes:

1. Patrick Flume: Welcomed guest attendees; introduced NORD Policy Manager Carolyn Sheridan
2. Carolyn gave NORD SC State Report Card Presentation
    a. Presentation provides evaluation of SC's effectiveness of serving the rare disease community within 9 target areas
        i. Presentation link will be made available on SC RDAC website
    c. Carolyn encourages members to attend April 25 Region C Policy & Advocacy Taskforce virtual meeting
    d. Members can share the NORD SC State Report Card with elected officials by clicking on button in presentation pdf
    e. SC legislation determines financial and health care policies that impact the rare disease community (and NORD's report card evaluation)
        i. Dr. Flume would like to see where other states had success in these areas and discuss in a following meeting where to prioritize our efforts
        ii. Carolyn will send Kendall copies of laws from nearby states
3. Rare Disease Day Symposium Debrief
    a. Inaugural symposium was held February 29 at Segra Park to coincide with Rare Disease Day; 26 in-person attendees, 47 virtual attendees
    b. Dr. Flume is pleased overall with the event turnout and balance of research and advocacy; open to suggestions for different locations and panel sessions for next year's event; would like to see more statewide involvement
    c. Evaluation Feedback
        i. Kendall provided feedback from 17 evaluation survey respondents collected so far; overall positive feedback
            1. Suggested Future Topics: patient panel; more information on clinical trials & statewide resources; more information on basic & translational research; speakers from diverse healthcare professions & roles they play in patient care; future treatment studies & curative therapies.
    d. Council feedback
        i. Guest attendee Sue Martin: attended symposium and found it valuable; would like higher attendance in future
        ii. Divya Ahuja: a breakdown of the audience would be helpful in determining who attended as either a peer or relative or fellow advocate; consider hosting in an appealing region like Charleston next year for bigger draw
        iii. Sue Martin: having a central location and keeping it affordable will help draw a larger audience; hosting it in Columbia can help draw elected officials
        iv. Divya Ahuja: SC Telehealth Association hosts an annual conference in February or March he helps organize; SC RDAC could have a table there in future
4. Other council projects
    a. Needs Assessment survey

        i. Here is the survey link: https://redcap.musc.edu/surveys/?s=PKMDXE3AX3EFJEWT
        ii. Kendall: currently 35 of the 61 recorded respondents have completed the survey; too few for thorough data analysis; a summary of findings will be available on the SC RDAC website once analyzed
        iii. Assessment of why not all respondents completed the survey, or why such few numbers: literacy rates, rural areas, lack of resource accessibility for knowledge of survey
        iv. Dr. Flume is seeking ideas for further dissemination
            1. Carolyn Sheridan has offered to share it on NORD
            2. Jonathan Hawayek: other states have used NORD's patient advocacy group list to disseminate surveys
            3. Sue Martin: has disseminated through her BDA newsletter and social media pages; suggests making a patient story or video about the survey's importance
            4. Taylor Dominguez: can share with RDDC and SC advocacy members; has offered to translate the survey into Spanish for Hispanic population
            5. Ginie Daguise: asks about its readability; Kendall says it was compiled for a lay public audience, but can review it
            6. Dr. Flume: survey was modified from other states' surveys; will reach out to contact in Colorado RDAC re: their survey responses for a benchmark

5. Housekeeping and Additional Topics
    a. Thoughts on replacing council member (Rare Disease Organization)
        i. Dr. Flume requests that council members send suggestions
        ii. Carolyn Sheridan offered to push notice of vacancy to patient advocacy groups
    b. SC Rare Disease website (scraredisease.org)
        i. Kendall is presenting a vision for website changes to Dr. Flume next week
        ii. updates will take time due to gap in training level and cooperation with website management team; welcomes council feedback as updates are made
        iii. Events page was added to site and Symposium event details and Agenda pdf were posted on it; landing page has today's meeting and link posted
    c. Old Business
        i. Dr. Flume: requested that Jon Hawayek submit list of top 50 rare diseases from Florida and Ginie Daguise submit from SC; plan is to obtain state data analytics
            1. Jon Hawayek: SPARK had commissioned outside consultant in Florida to provide top 50 of rare disease prevalence and provide ICD10 code and NDC code for treatment; now working on top 51-500 of rare disease prevalence

            2. Ginie Daguise: does not have access to data sets for SC; suggests contacting Dr. Anna Lopez -De Fede of Family Medicine Institute as she works with Medicaid data and could facilitate research request
        ii. Jon Hawayek: most state employee health plans use PBM; Florida requested through PBM to pull data from state employee health plans
        iii. Tripp Jennings: data would be available through RFA or revenue & fiscal affairs
        iv. Ginie Daguise: a number of programs provide funding, looking into possible costs
        v. Kate Wink: thought state agencies could only charge for copies; not the case
        vi. Dr. Flume: we could compile the data and then submit an analysis request and see if it's an allowable expense
    d. Please send updated CVs and headshots to Kendall (spillman@musc.edu)
    e. Kendall will send NORD State Report Card Presentation with meeting minutes in forthcoming email and add to website
    f. Schedule next meeting
        i. Kendall will be in touch to schedule a quarterly meeting for July
6. Adjourn

SC Rare Disease Advisory Council: Mission and Responsibilities

Mission: The council shall advise the Governor, the General Assembly, and other stakeholders on research, diagnosis, treatment, and education related to rare diseases

Definition: For purposes of this council, a rare disease is defined as one affecting fewer than 200,000 persons combined in a particular rare disease group

Responsibilities of the Advisory Council:

- Solicit comments from stakeholders, including patients and patient caregivers in South Carolina impacted by rare diseases, to assess the needs of rare-disease patients, caregivers, and providers in the State;
- Consult with experts on rare diseases to develop recommendations to improve patient access to and quality of rare-disease specialists, affordable and comprehensive health care coverage, relevant diagnostics, timely treatment, and other needed services;
- Research and identify priorities related to treatments and services provided to persons with rare diseases in South Carolina and develop recommendations that include safeguards against discrimination for these populations on such issues, including disaster and public health emergency-related planning;
- Publish a list of existing, publicly accessible resources on research, diagnosis, treatment, and education relating to the rare diseases in South Carolina;
- Identify and distribute educational resources to foster recognition and optimize treatment of rare diseases in South Carolina; and
- Identify best practices to reduce health disparities and achieve health equity in the research, diagnosis, and treatment of rare diseases in South Carolina.

- Report annually (by June 30) to the Governor, the Chairman of the Senate Finance Committee, the Chairman of the Senate Medical Affairs Committee, the Chairman of the House Ways and Means Committee, and the Chairman of the House Medical, Military, Public and Municipal Affairs Committee.