

Rare Diseases Advisory Council

January 5, 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)	X
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	
South Carolina Hospital Association	Kate Wink (Santee Cooper)	X
South Carolina Primary Healthcare Association	Vicki Young (SCPHCA)	
Biopharma Industry	Jonathan Hawayek (SPARK Therapeutics)	
Research and Treatment of Rare Diseases (3)	Maysen Mesaros, MS (MUSC, neuroscience)	X
	Neena Champaigne, MD (Pediatric Genetics)	X
	Chip Norris, PhD (MUSC, connective tissue)	
Patient (2)	Anthony Hamilton (familial TTR amyloid)	
	Karen Kemper, PhD (scleroderma)	X
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
<u>Guests In Attendance:</u>		
MUSC - SCTR Success Center	Stephanie Gentilin (Director)	X
	Tara Pittman (Recruitment Manager)	X
Genentech	Sonya Rigsby (Therapeutic Area Manager)	X

Meeting Minutes:

1. Patrick Flume: Welcome & reminder of state funding approval
 - a. Contract between MUSC and MUHA finalized; funding fairly restrictive to what we can do.
 - b. Allocated \$250,000 to use; cannot pay ourselves and likely cannot pay for travel.
2. Introductions
 - a. Kendall Spillman introduced as new RDAC Program Coordinator

3. Rare Disease Day Symposium

- a. Venue- Segra Park Club Lounge February 29, 2024, 9:30 am-2 pm (1640 Freed Street, Columbia, SC 29201)
 - i. This is inaugural event; held on last day of February, this year being Leap Day, in conjunction with National Rare Disease Day
 - ii. Venue contract will include A/V support and catered boxed lunch
- b. Attendees
 - i. Council feels this audience will likely be members of the public, more so than the scientific community
 1. Should still be scholarly enough for scientists to recognize potential interest areas or collaborations
 - ii. Event will be free and hybrid with livestream for public access
 1. Registration will be encouraged. Registration link to be forwarded to council when ready
 2. Council generally in favor of having event recorded. Will look into speaker consent for, and audience acknowledgement of, recording.
 - iii. Invitations for legislature
 1. They'll be in session on weekday, so might not be able to attend
 2. Last year's event was geared towards them, so it should be fine to have a different focus
- c. Agenda
 - i. Divided between research and advocacy (2 hours for each)
 1. Initially thinking each speaker will be given 20 minutes to present, with 10-minute Q&A session, but this is not fixed
 2. Must be non-MUSC centric. Should include speakers from across the state.
 3. Council in agreement that a patient panel can be impactful for the public
 - ii. Potential agenda theme and outline:
 1. General Info on Clinical Trials & Patient Engagement
 - a. Presented by Dr. Flume?
 - b. Discuss the value of clinical trial participation, overcoming concerns related to genetic research, resources for getting involved in research (ClinicalTrials.gov, SCresearch.org, Researchmatch.org), etc.
 2. Innovation and Discovery happening within South Carolina
 - a. Genetics and Rare Disease
 - i. Dr. Judge to discuss In Our DNA?
 - ii. DHEC overview of state-wide genetics testing efforts?
 - iii. Perhaps someone from Clemson to discuss their genetics program?
 - iv. Newborn Screening to be discussed by Neena Champaigne

1. Data to be shared to aid in discussion from SCDHEC?
 - v. Someone from Greenwood could walk through animal models / functional studies?
 - b. Diseases focused presentation?
 - i. Chip Norris to present on Ehlers Danlos Syndrome research
 3. Patient Experience and Advocacy
 - a. Consider inviting patients to speak who also have a role in or experience with advocacy work, so they can speak to both personal experience and advocacy efforts (3-4 person panel)
 - d. Evaluation
 - i. Council to debrief post-symposium
 - ii. Post-event survey to be distributed to attendees for feedback
4. Other council projects
 - a. Needs Assessment survey
 - i. Here is the survey link:
<https://redcap.musc.edu/surveys/?s=PKMDXE3AX3EFJEW7>
 - ii. Council can begin distribution immediately.
 - iii. Please email Kendall with where/to what groups the link has been distributed so we can track dissemination and not duplicate efforts**
 - iv. Karen Kemper to share information on and disseminate the needs assessment at the state-wide Patient Education Conference on April 13th
 - v. Other messaging and marketing materials to help further promote the needs assessment is forthcoming
 - vi. Ideas for dissemination?
 - b. SC Rare Disease website (scrareddisease.org)
 - i. Working with MUSC web team for redesign. Open for content suggestions/ideas from council
 - ii. Idea to create Resources link for patients and families
 - c. Contact Registry
 - i. Discussion item to be followed up on post-symposium
 1. Follow up on Medicaid database utilization needed
 - d. SCTR to work with science writing intern to promote RDAC activities; Kendall and Tara will meet with her next week to discuss dissemination
 - e. RDAC will share a table with SCTR at the MUSC Senior Expo on March 13
5. Housekeeping and Additional Topics
 - a. Please send updated CVs and headshots to Kendall (spillman@musc.edu) by 02/01/24. Kendall to send reminder email
 - b. Swag representing SCTR and SCResearch.org will be ordered for symposium
 - i. Ordering items from preferred vendor Concorde LLC [Marianne Thomason, Mt. Pleasant | Promo - Home \(concordesc.com\)](http://MarianneThomason.Mt.Pleasant|Promo-Home.concordesc.com). Send any suggestions to Kendall (we will need to order in the next week or two)
 - c. Schedule next meeting

- i. Kendall will be in touch to schedule a quarterly meeting for April
6. Resources
 - a. Reminder of Rare Disease Clinical Research Network website & newsletter [Subscribe to the Spotlight Newsletter | Rare Diseases Clinical Research Network \(rarediseasesnetwork.org\)](#)
 - b. Rare Disease Research Webinar to be held January 18. Registration link provided [How Gene Therapy Will Move Rare Disease Research at Warp Speed \(advarra.com\)](#)
7. 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.