

## Rare Diseases Advisory Council Quarterly Meeting

October 3, 2025

9:00 AM-10:00 AM

### Meeting agenda:

- Welcome
- The Role of the RDAC in Providing Recommendations to the State, Mr. Mark Sweatman

Mark clarified that while providing factual information is appropriate, offering recommendations—such as commenting on legislation—requires careful consideration.

The group considered whether to proactively request opportunities to speak on relevant bills. Mark and Kate offered to monitor bills and keywords on the follow list and notify the committee when subcommittee comments are needed. The group emphasized the importance of maintaining the committee's focus on rare diseases, and discussed identifying bills where RDAC could testify, nominating speakers, and ensuring they are well-prepared to represent the council.

The group inquired about other councils with similar structures, and Mark agreed to review existing provisos to identify comparable setups. The conversation also touched on the issue of carryover funds. Mark supported adding language to support fund carryover and will connect with Patrick and Tara to refine the language.

- Planning for the 3<sup>rd</sup> Annual Rare Disease Symposium – Discussion
  - In conversation with Segra Park (same venue as previous years)
  - Tara met with Segra Park's new AV company who was very responsive to past concerns. Proposed improvements:
    - Use of TVs and wall projection (not windows)
    - New camera and microphone setup
    - Buffet-style lunch (including vegan/vegetarian salad options)
  - City of Columbia has designated the area as a paid parking zone. Tara to inquire about parking validation options.
  - Legislative Attendance Consideration - Friday is not ideal for inviting legislators or executive branch members (in session Tuesday–Thursday). Consider audience when planning date and outreach.
  - Form a working group to develop agenda and presentation themes
    - Participants: Karen K., Neena C., Jon H., Patrick F.
    - Group to meet after the NORD event (before end of October)
  - Speaker & Topic Suggestions
    - Open call for topic ideas and speaker recommendations
    - Mention of Palmetto Health Collective as a potential contributor
    - Consider highlighting report content (e.g., enteral nutrition) with patient stories
- Other Updates
  - NORD Rare Action on the Road Event & Other Collaborations

- Dr. Champaigne presented to a group of 20 attendees about the RDAC and our recent annual report.
- Key takeaway - there are a lot of groups across the state focusing on similar things. Important to find ways to connect and collaborate.
  - [Palmetto Health Collective](#)
  - [SC Rare](#) (Carol Shea Linton, CEO, present on the meeting today – group to connect further)
  - [SAIM Coalition](#)
  - South Carolina Hemophilia and Bleeding Disorders Coalition
- RD Data
  - Dr. Kit Simpson is continuing her work on RD prevalence data. The group discussed Epic Cosmos data as a source of clinical data (as opposed to claims data). Dr. Flume discussed potential opportunities to statewide partnerships (e.g. USC, DPH, etc.) to better understand and query the data. The council to send names of potential collaborators to Dr. Flume to explore further.
- Adjourn

**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

**Membership:**

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	X
Department of Public Health	Ginie Daguise (SCDPH)	X
Department of Health and Human Services	Kevin Wessinger (SCDHHS)	X
Greenwood Genetics	Steve Skinner, MD (President & CEO)	
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)	
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)	X
Patient (2)	Karen Kemper, PhD (scleroderma) Bridget Downing	X X

Rare Disease Organization	Yvonne Donald, M.A., CSCEC (James R. Clark Memorial Sickle Cell Foundation)	
Caregiver of person with rare disease	Cara O'Neill, MD, FAAP (Cure Sanfilippo Foundation)	
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	
Program Coordinator	Tara Pittman Kimberly Brown	X X

**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.