

**Rare Diseases Advisory Council January Quarterly Meeting**  
**Meeting Minutes**

January 10<sup>th</sup>, 2025

9:00 AM-10:00 AM

**1. Welcome & Introductions**

- a. Dr. Steve Skinner, Greenwood Genetics President & CEO
- b. Bridget Downing, Patient

**2. Rare Disease Symposium Plans | February 28<sup>th</sup> @ Segra Park (Columbia, SC)**

a. *Draft Agenda*

<b>Time</b>	<b>Title/topic</b>	<b>Speaker</b>
9:30	Check In/ Registration	
10:00	Welcome/opening remarks	Dr. Patrick Flume
10:10	State of the state <ul style="list-style-type: none"> <li>• Show data (Ginie Daguise) and/ or report card</li> </ul>	<i>Jonathan Hawayek to reach out to Carolyn Sheridan from NORD for availability</i>
10:30	Changing policy in South Carolina <ul style="list-style-type: none"> <li>• Approaches to Rare Disease in Legislation, Challenges in Legislation</li> </ul>	Ken Bingham or Emily Bingham <i>Follow up on availability from Jonathan Hawayek</i>
11:00	Discovery/translational science <ul style="list-style-type: none"> <li>• Zebrafish</li> </ul>	Greenwood Genetics Mike Lyons or Heather Rich <i>Follow up on availability from Steve Skinner</i>
11:45	Networking Lunch	
12:00	Personal story (led to doing science)	Tara O'Sullivan <i>Follow up on availability from Neena Champaigne</i>
12:30	Personal story: Participation in clinical trial	Cara O'neal <i>Follow up on availability from Neena Champaigne</i>
1:00	Q&A for Personal Stories	
1:30	Advocacy development <ul style="list-style-type: none"> <li>• FDA/critical path institute</li> </ul>	Smith Hevner <i>Follow up on availability from Karen Kemper</i>
1:50	Closing remarks	

- b. *Speaker/ Presenter Ideas*
  - i. *FDA/ Critical Path Institute Presentation Details: Drug Repurposing*
    1. Identified as a potential topic for the symposium and future Council focus.
    2. Emphasis on highlighting resources and examples of successful repurposing efforts.
- c. *Actions:*
  - i. Titles for agenda items to be revised to appeal to audience
  - ii. Potential adjustments to time allocations (e.g., Q&A sessions, networking lunch).
  - iii. Verify speaker availability
  - iv. Promotion
    1. Halley & Tara meeting with P&C

### **3. Housekeeping and Additional Topics**

- a. *SC RDAC Charter Update*
  - i. Charter was amended based on feedback from Mark Sweatman
  - ii. **Next Steps:**
    1. Distribute updated Charter for review and electronic vote.
    2. Voting and finalization to be completed within two weeks.
    3. Finalized Charter to be posted on the RDAC website.
- b. *Needs Assessment Promotion*
  - i. Distribution through advocacy groups and MUSC inpatient TVs.
  - ii. Recent uptick in responses
  - iii. Materials and links available for dissemination by Council members
    1. Sent to Sonya & Dr. Steve Skinner
- c. *Follow Ups from Last Meeting*
  - i. Jonathan Hawayek sent full data set to Ginie Daguise
    1. Challenges Identified:
      - a. Small sample sizes make mapping rare diseases difficult.
      - b. Need for more comprehensive data from state health plans and private payers.
    2. Next Steps:
      - a. Compile and refine a list of specific data requests.
      - b. Ensure data aligns with the national incidence to maintain quality checks
      - c. Follow up with Ginie on progress/ findings

### **4. Adjourn**

**Action Items**

1. **Charter Review and Vote**
  - Updated Charter to be distributed for feedback and approval.
2. **Data Analysis**
  - Members submit specific data needs to Dr. Daguise.
3. **Symposium Planning:**
  - Confirm speakers, refine agenda titles and time frames
  - Finalize promotional strategies and distribute registration links.

**Membership:**

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	P
Department of Health and Environmental Control	Ginie Daguise (SCDHEC)	P
Department of Health and Human Services	Kevin Wessinger (SCDHHS)	A
Medical University of South Carolina	Dan Judge, MD (MUSC, cardiology)	A
Greenwood Genetics	Steve Skinner, PhD (Greenwood Genetics)	P
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	A
South Carolina Hospital Association	Kate Wink (Santee Cooper)	A
South Carolina Primary Healthcare Association	Vicki Young (SCPHCA)	A
Biopharma Industry	Jonathan Hawayek (SPARK Therapeutics)	P
Research and Treatment of Rare Diseases (3)	Maysen Mesaros, MS (MUSC, neuroscience)	P
	Neena Champaigne, MD (Pediatric Genetics)	P
	Chip Norris, PhD (MUSC, connective tissue)	
Patient (2)	Karen Kemper, PhD (scleroderma)	P
	Bridget Downing	P
Rare Disease Organization		P
Caregiver of person with rare disease		
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	A
Program Coordinator	Halley Harris	P
	Tara Pittman	P

Other Attendees: Stephanie Gentilin, Sonya Rigsby, Russell Spencer- Smith

**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.