Rare Diseases Advisory Council Quarterly Meeting

July 11th, 2025

9:00 AM-10:00 AM

Meeting agenda:

- Welcome & Updates
    - The annual report was submitted to the Governor's office and will be posted to the RDAC website soon.

- Estimation of Rare Disease Prevalence in South Carolina, Dr. Kit Simpson
    - Dr. Simpson presented a pilot study on estimating rare disease prevalence in South Carolina. She described the methodology using multiple datasets and a modified Peterson capture-recapture method. Ten rare diseases were analyzed using data from Medicare, MarketScan, and the SC Revenue and Fiscal Affairs Office (RFA). Key findings included prevalence estimates and demographic breakdowns. Limitations and future improvements were discussed, including the need for Medicaid data and validation of ICD-10 code groupings.
    - Members provided feedback on data discrepancies and the importance of accurate representation. Suggestions included using Medicaid data for pediatric conditions and grouping disorders for better service planning. The need for collaboration with patient advocacy groups was emphasized.

- Administrative Updates
    - Discussion on council member term limits and succession planning.
    - Reminder of the upcoming Newborn Screening Advisory Committee meeting in August.
    - Announcement of the NORD Summit in October and delegate participation.
    - Preliminary discussion on hosting the symposium in February. Suggestions to form a subcommittee for planning and venue selection.

- 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)	A
Department of Public Health	Ginie Daguise (SCDPH)	A
Department of Health and Human Services	Kevin Wessinger (SCDHHS)	P
Greenwood Genetics	Steve Skinner, MD (President & CEO)	P
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	A
South Carolina Hospital Association	Kate Wink (Santee Cooper)	P
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) Neena Champaigne, MD (Pediatric Genetics) Chip Norris, PhD (MUSC, connective tissue)	P P A
Patient (2)	Karen Kemper, PhD (scleroderma) Bridget Downing	P P
Rare Disease Organization	Yvonne Donald, M.A., CSCEC (James R. Clark Memorial Sickle Cell Foundation)	A
Caregiver of person with rare disease	Cara O'Neill, MD, FAAP (Cure Sanfilippo Foundation)	P
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	P
Program Coordinator	Tara Pittman Kimberly Brown	P P

Non-member attendance: Kit Simpson, Jenna Doerr, Sonya Rigsby, Daphne Ni, Michelle Myer, Ingrid Ma, Stephanie Gentilin

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.