South Carolina Rare Disease Advisory Council

Rare Diseases in South Carolina

2024-2025 Annual Report

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Henry McMaster, Governor of South Carolina
Harvey S. Peeler, Jr, Chairman, Senate Finance Committee
Daniel B. Verdin, III, Chairman, Senate Medical Affairs Committee
Bruce W. Bannister, Chairman, House Ways and Means Committee
Sylleste H. Davis, Chairman, House Medical, Military, Public and Municipal Affairs Committee

June 24, 2025

Re: Progress report for the SC Rare Disease Advisory Council

Dear Governor and respective Chairmen,

On behalf of the members of the South Carolina Rare Disease Advisory Council, it is my pleasure to submit the second annual legislative report as outlined in Section 44-1-320 of Bill H.3956. This report documents the key initiatives related to research, advocacy, and awareness that the council sought to achieve over the past year. Please do not hesitate to reach out with any questions or comments.

Sincerely,

Patrick A Flume, M.D.
The Powers-Huggins Endowed Chair for Cystic Fibrosis
Distinguished Professor of Medicine and Pediatrics
Chair, SC Rare Disease Advisory Council
Director, MUSC Cystic Fibrosis Center
Associate Vice President for Clinical Research, MUSC

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1. Executive Summary

1a. Background of the council

The South Carolina Rare Disease Advisory Council (SC RDAC) was originally established by law in 2021 (Section 44-1-320 of Bill H. 3956) to address the needs of those living with rare diseases. There are currently 29 states that have enacted legislation to establish an RDAC.[1] The South Carolina General Assembly through Proviso 117.146(E)(GP: Rare Disease Advisory Council) of the 2024-2025 South Carolina Appropriation Act authorized the South Carolina Department of Health and Human Services to continue funding to the Medical University of South Carolina to house and provide coordination and management of the SC RDAC. The council advises the Governor, the General Assembly, and other stakeholders on research, diagnosis, treatment, and education relating to rare diseases. This year, as the proviso outlined, a representative from Greenwood Genetics was added to the council's membership.

1b. Summary of council initiatives

Since the last progress report submitted in June 2024, the council has tackled several key initiatives as outlined in its official responsibilities. Main accomplishments of the council during the 2024-2025 year are highlighted below and will be discussed in greater length throughout the report:

- Added the President and CEO of Greenwood Genetics to the council (per the most recent proviso), in addition to the replacement of previous members representing the rare disease patient and caregiver populations;
- Created and voted to approve a charter to establish clear guidelines and expectations terms of service for the council;
- Held quarterly meetings open to the public to encourage statewide collaboration on, and support of, council initiatives;
- Hosted the second annual Rare Disease Symposium on Rare Disease Day (February 28th) to promote research and spread awareness of and advocacy for the state's rare disease community (and had two local stadiums light up venues in rare disease colors in recognition of Rare Disease Day);
- Completed data collection for the needs assessment survey created and released in 2024;
- Piloted a study to obtain the first statewide estimates of rare disease prevalence in South Carolina, using hospital and insurance data to provide a more complete picture of who is affected; and,
- Maintained the official website (rarediseasesc.org) to optimize awareness of state resources and foster public engagement

[1] National Organization for Rare Disorders, https://rarediseases.org/policy-issues/rare-disease-advisory-councils/

The council reviewed the most current Rare Disease State Report Card for South Carolina developed by the National Organization of Rare Diseases (NORD) and compared it not only to South Carolina's previous grades, but also those of comparable states. Although the state scores poorly in several areas, the council will, in this report, call attention to proposed state legislation currently under review that addresses some of these topics.

Council member, Dr. Neena Champaigne, attended NORD's 2nd annual RDAC leadership meeting on behalf of the SC RDAC, held in Washington, DC. There was representation from RDACs across the nation at this event. The day included networking, panel sessions on state budgets, prescription drug affordability boards and the FDA, as well as breakout sessions on surveys, Medicaid, policy, and patient engagement. Dr. Champaigne then attended the subsequent NORD's 2-day Rare Disease and Orphan Products Breakthrough Summit; the costs for this summit were covered by NORD.

The council's mission, membership, and official responsibilities are also provided to restate the council's purpose and role as a statewide project.

Finally, the minutes from the quarterly meetings of the SC RDAC and the expense report follow in this progress report.

2. RDAC mission and membership

2a. Background

A rare disease is defined by the Orphan Drug Act as a disease or condition that affects less than 200,000 people in the country.[2] According to the U.S. Food & Drug Administration (FDA), more than 30 million people in the US are affected by more than 7,000 rare diseases.[3] Of these known rare diseases, approximately 95% have no treatment.[4] Not only does this significantly impact the quality of life and access to adequate care for those living with a rare condition, but it also poses a significant economic burden on society. Supported by IQVIA (a company that provides data and technology solutions for the healthcare industry), Chiesi Global Rare Diseases (a rare disease therapeutics company) studied the direct, indirect and mortality costs associated with a sample of rare diseases and compared these costs to those of mass market diseases. They found that, on a per patient per year basis, the rare disease economic burden is approximately 10x higher than that of mass market diseases (and this is before factoring in social costs, including the impact on a patient's health related quality of life).[5] Available treatments for rare conditions help to reduce some of that additional burden. This evaluation is just one example of the value of supporting the development of rare disease therapies, diagnostic tools and newborn screenings. The SC RDAC allows government to better

[2] U.S. Food & Drug Administration, 2018, https://www.fda.gov/industry/designating-orphan-product-drugs-and-biological-products/orphan-drug-act-relevant-excerpts
[3] U.S. Food & Drug Administration, 2024, https://www.fda.gov/patients/rare-diseases-fda
[4] National Organization for Rare Disorders, 2019, https://rarediseases.org/wp-content/uploads/2019/01/RDD-FAQ-2019.pdf
[5] Chiesi, 2022, https://chiesirarediseases.com/assets/pdf/chiesiglobalrarediseases.whitepaper-feb.-2022_production-proof.pdf

understand the impact of rare disease at both an individual and state level so informed and impactful legislative decisions can be made.

2b. Mission

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

2c. Membership

The council is currently composed of seventeen members and intended to be broadly representative of stakeholders. Each position is appointed by statewide leadership including the President of the Medical University of South Carolina (MUSC), the Dean of the University of South Carolina (UofSC) School of Medicine, the Director of the Department of Health and Environmental Control (DHEC), the Director of the Department of Health and Human Services (DHHS), the Executive Director of the South Carolina Hospital Association (SCHA), the Executive Director of the South Carolina Primary Healthcare Association (SCPHA), the Executive Director of the State Public Benefit Authority (SPBA) and the Director of Greenwood Genetics.

Representation	Member
Medical University of South Carolina	Patrick Flume, MD Council Chair
Department of Public Health	Virginie Daguise, MSPH, PhD
Department of Health and Human Services	Kevin Wessinger, MD
Medical University of South Carolina	Dan Judge, MD (cardiovascular genetics)
Greenwood Genetics	Steve Skinner, MD
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (infectious disease)
South Carolina Hospital Association	Kate Wink (Santee Cooper)
South Carolina Primary Healthcare Association	Vicki Young
Biopharma Industry	Jonathan Hawayek (SPARK Therapeutics)
Research and Treatment of Rare Diseases (3)	Maysen Mesaros, MS (neuroscience) Neena Champaigne, MD (pediatric genetics) Chip Norris, PhD (connective tissue)
Patient (2)	Karen Kemper, PhD (scleroderma) Bridget Downing (Friedreich's ataxia)
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 (BCBS South Carolina)

2d. 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 an initial list and an updated 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 prevention, recognition and optimization of treatment of rare diseases in South Carolina.
- Identify best and effective practices to reduce health disparities and achieve health equity in the research, diagnosis, and treatment of rare diseases in South Carolina; and
- Create and maintain a current public website that shall include the annual reports, meetings notices and minutes, and generally related resources as well as those developed from its research efforts. The SCRDC members shall serve without compensation and/or per diem.
- Provide an annual report entitled South Carolina Rare Disease Council Annual Report (SCRDCAR) no later than June 30, 2025, 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. The SCRDCAR shall describe the activities and progress of the SCRDC and provide recommendations to the Governor and General Assembly on ways to address the needs of people living with rare diseases in the state of South Carolina.

2e. Council activities and operations

In the past year, the council has had quarterly virtual meetings (October 2024, January 2025, April 2025). These meetings have been open to the public (with the Microsoft Teams links available on the SC RDAC website, rarediseaseSC.org). In addition to these quarterly meetings, subgroups convened to plan the South Carolina Rare Disease Symposium. A council member represented the SC RDAC at both NORD's 2nd annual in-person RDAC leadership meeting as well as its 2-day Rare Disease and Orphan Products Breakthrough Summit. In May 2025, several RDAC members began attending the Newborn Screening Advisory Committee quarterly meetings.

New to the council this year was the establishment of a charter, outlining the requirements and expectations of membership, terms of service and decision-making processes.

3. Council projects and achievements

3a. Rare Disease Day and the 2nd Annual South Carolina Rare Disease Symposium

The council held its 2nd annual Rare Disease Symposium on February 28, 2025, coinciding with the Rare Disease Day. The purpose of the symposium was to promote research, advocacy, and awareness around rare diseases in South Carolina. The catered, hybrid event was held at Segra Park in Columbia, SC from 10 am-2 pm, with 70 registered to attend in person and 100 virtually (an increase of over 70 registrants from the previous year). Promotion of the event included print and digital advertisements placed via The Post and Courier (the largest newspaper in the state with significant reach both in print and online) to help increase reach to individuals from across the state. Ad placement locations included greater Charleston, Columbia, Beaufort County, Myrtle Beach, Greenville/Spartanburg, and Florence (Appendix A).

The reaction from attendees regarding the event was overwhelmingly positive. A few anonymous remarks left on the event feedback survey include:

- “This was one of the best events I've been to, especially considering its relatively small size. Nothing felt like an afterthought or an outlier, every session felt like a critical part of the whole day.”
- “Its impressive to be educated in legal/political development, the latest in research, healthcare models, resources for assistance, medical information, etc. all in a 4 hour symposium!”
- “It was both detailed and expansive, capturing a wide gamut of the causes and effects and deep impacts rare diseases have on patients, families, communities, research scientists, and healthcare providers.”
- “This symposium provided a platform to hear from and interact with researchers, patients, physicians and local government with the common goal of improving outcomes for rare disease patients. As a researcher who is new to SC this was an excellent opportunity to network with local rare disease community.”

To bring additional attention to Rare Disease Day, the council solicited the support of two local venues to participate in the NORD-initiated “Light Up for Rare” campaign, during which local stadiums and landmarks temporarily illuminate their structure in colors representing rare disease awareness. Both the Jospeh P. Riley Jr. Park (downtown Charleston, SC) and Credit One Stadium (Daniel Island, SC) participated during the evening hours of February 28th.

3b. Rare Disease Needs Assessment

In early 2024, the council disseminated a needs assessment survey for patients and caregivers of patients with rare diseases in South Carolina. The survey was closed in Spring 2025 after it was completed by 82 people representing a wide range of conditions, ages, and geographic locations.

The needs assessment identified key areas for improving care for rare disease patients in South Carolina. Top priorities include better access to affordable care, such as travel support, medication cost relief, and expanded insurance coverage. Respondents called for increased education for both providers and patients, improved access to specialists, and stronger support systems for families and advocacy. Additional needs include more research funding and broader healthcare system improvements, such as comprehensive care models and expanded telehealth services. Greater detail is provided in Appendix B.

3c. South Carolina Rare Disease Incidence Data

South Carolina currently lacks reliable data on how many residents are living with rare diseases—a gap that limits the state's ability to plan and allocate healthcare resources effectively. To address this, researchers conducted a pilot study to estimate the prevalence of rare diseases using available state and national data sources (Appendix C). This work lays the foundation for a scalable, cost-effective method to support evidence-based policy decisions.

The study combined two key data sources from 2022: South Carolina's All-Payer Claims Database (RFA), which includes hospital, emergency room, and outpatient surgery records, and a national 5% Medicare sample, which captures office-based care not included in the RFA data. This dual-source approach allowed researchers to estimate how many patients were missing from hospital-based data alone, particularly those who only received care in physician offices.

The report includes estimates for ten rare diseases, showing both the number of patients found in hospital and surgery center records (Table 1, Column 1), and the adjusted numbers that also account for people who only visited a doctor's office (Table 1, Column 2). These adjusted numbers give a more complete picture of how many people in South Carolina were living with each condition in 2022. The study also provides demographic breakdowns by age, gender, and race, offering valuable insights for targeted interventions.

Table 1: Results for 10 Rare Disease Estimated Using the SC All-Payer Data Set for 2022 with Adjustment for Missing Cases with Only Medical Office Visits in the Year.

Rare Disease Name	Observed Cases	Estimated Cases	Age Median (range)	Percent Female	Percent White	Percent Black	Percent Other
Polycystic Kidney Disease	731	928	53 (1-85+)	54%	56%	38%	6%
Sjogren's Syndrome	4,452	6,811	60 (1-85+)	93%	74%	22%	4%
Scleroderma	1,888	3,055	59 (4-85+)	39%	68%	29%	3%
Polycythemia Vera	1,277	2,363	63 (3-85+)	39%	86%	12%	2%
α 1 Antitrypsin Deficiency	660	900	63 (2-85+)	57%	94%	4%	2%
Duchenne Muscular Dystrophy	127	209	18 (1-70)	15%	72%	16%	2%
Von Willebrand's Disease	997	1,274	38 (1-85+)	78%	82%	13%	5%
Cystic Fibrosis	793	793	22 (1-85+)	57%	81%	14%	5%
Amyotrophic Lateral Sclerosis	671	1,063	62 (19-85+)	44%	76%	20%	4%
Guillain Barre Syndrome	1,477	1,618	57 (3-85+)	52%	76%	20%	4%

Importantly, the research team developed and validated a repeatable method for estimating rare disease prevalence using existing data infrastructure. Although the pilot study was to validate the method on 10 rare diseases, this approach can be applied to many more conditions with minimal additional cost. The team recommends updating the analysis with 2023 data to confirm the stability of adjustment factors and streamline future estimates. Once validated, this method could be used efficiently to estimate prevalence for up to 100 rare diseases.

For policymakers, this study offers a practical roadmap to improve rare disease surveillance in South Carolina. With better data, the state can more effectively plan services, allocate funding, and advocate for the needs of individuals and families affected by rare conditions. Continued investment in this data-driven approach will ensure that South Carolina remains proactive in its healthcare planning.

4. Current issues under legislative review

The council has consulted with patients, caregivers, healthcare providers, and advocacy groups to better understand the ongoing challenges faced by the rare disease community. Through this engagement, the council has identified three key policy issues currently under legislative review that address health outcomes and access to care across South Carolina.

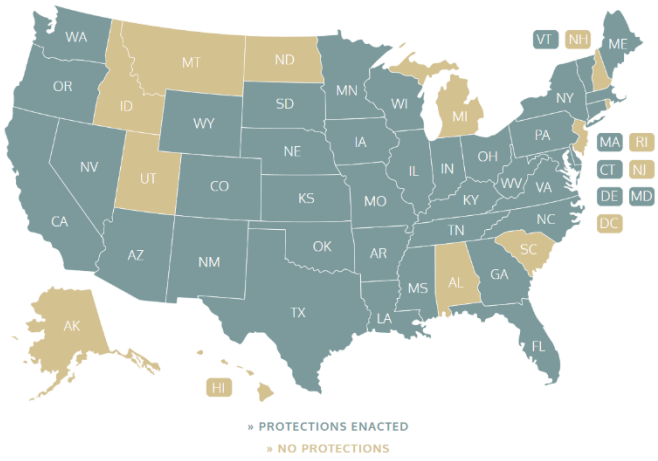
4a. Copay Accumulator Adjustments

Rare disease treatments often come at high out-of-pocket cost, and many patients rely on manufacturer copay assistance to afford their medications. When insurers use copay accumulator programs, this assistance does not count toward a patient's deductible or out-of-pocket maximum, often leading to sudden, unaffordable expenses. More than 20 states have enacted laws limiting copay adjustment programs, including Georgia, Tennessee, and North Carolina.[6] The council notes there is legislation introduced in the 126th Session of the South Carolina General Assembly addressing this issue (H. 3934 and S. 330).

4b. Step Therapy Protocols

Step therapy, also called "fail first," is a practice used by insurance companies that requires patients to try alternative, usually less expensive, medications before they can get the one their doctor originally prescribed. Although this approach is meant to help control healthcare costs, it is often applied without considering a patient's individual medical history or the judgment of their doctor. This can lead to delayed access to medically necessary treatments, placing rare disease patients at unnecessary risk of progression and irreversible harm, and, in many cases, increase overall healthcare costs instead of reducing them.[7]

38 states have implemented step therapy reform laws that preserve clinician authority and protect patients from being forced to repeat ineffective or harmful treatments; at the same time, these protections allow insurance plans to manage costs effectively when doing so does not compromise patient care. The council notes that legislation that was introduced in the 126th Session of the South Carolina General Assembly (S. 531).



Step Therapy Legislation by State (2024)
https://steptherapy.com/step-therapy-legislation-by-state/

[6] All Copays Count, 2025, https://allcopayscount.org/state-legislation-against-copay-accumulators/
[7] Avalere Health, 2020, https://advisory.avalerehealth.com/insights/step-therapy-can-lead-to-higher-oop-costs-for-crohns-disease-patients

4c. Telehealth Access

Telehealth is a crucial resource for rare disease patients, reducing the need for extensive travel—often hundreds or thousands of miles—to access specialized care, diagnosis, and ongoing treatment. Data from the council's recently conducted needs assessment shows how significant this burden is:

- 39% of patients must travel greater than 50 miles in South Carolina to access care related to their rare disease.
- 40% of patients must travel out of state to access care related to their rare disease.
- One of the chief complaints leading to dissatisfaction related to accessing specialists and additional testing/diagnostics is travel burden.

Expanded access to telemedicine increases accessibility to healthcare, reduces healthcare costs, and improves patient outcomes. Broader implementation of cross-state licensure arrangements would ensure continued access to expert care regardless of geographic location. 41 states are part of the Interstate Medical Licensure Compact (IMLC), which streamlines the licensing process for physicians practicing in multiple states while maintaining a uniform and stringent standard. The Council notes that legislation was introduced in the 126th Session of the South Carolina General Assembly on this topic (H. 4335 and S. 377.

5. Expense Report

	FY2024 NCE 07/01/24-10/31/24	FY2025 11/01/24-6/31/25	Total
Salaries	$57,535	$92,678	$150,213
Fringe	$25,661	$41,335	$66,995
Symposium and Outreach	$10,498	$18,244	$28,742
Data Analytics	$0	$10,000	$10,000
Annual Report Postage	$113	$0	$113
Travel	$0	$4,034	$4,034
	$93,807	**$166,291**	**$260,098**

Appendix A. South Carolina Rare Disease Symposium Advertisements and Agenda

Sample Advertisements

Event Agenda

SC Rare Disease Symposium

9:30 Check in / Registration

10:00 Welcome & Opening Remarks

Patrick Flume, MD, Endowed Chair, Power-Huggins Endowed Chair for Cystic Fibrosis & Chair for the Rare Disease Advisory Council, Medical University of South Carolina (MUSC)

10:10 South Carolina's Rare Disease State Report Card

Carolyn Sheridan, MPH, State Policy Manager, Eastern Region, National Organization for Rare Disorders (NORD)

10:30 Effective Advocacy

Emily Keeney, Government Relations Advisor, Adams and Reese LLP

11:00 Journey of Discovery: An integrated model of care to find answers and develop treatments for rare genetic diseases

Dr. Mike Lyons, R. Boykin Curry Chair in Translational Genomics and Therapeutics; Director of Clinical Services; Senior Clinical Geneticist, Greenwood Genetics

Dr. Heather Flannagan- Steet, Director of Functional Studies, Greenwood Genetics

11:40 Journey of Discovery Q&A

11:50 Lunch

12:15 Personal Stories/Advocacy Presentations

Building Hope: A decade of supercharging research and advocacy for Sanfilippo syndrome

Glenn O'Neill, Founder & CEO of the Cure Sanflippo Foundation

The Other Side of Impossible

Tarah O'Sullivan*,* CEO and founder of the Drake Rayden Foundation

12:45 Personal Stories/ Advocacy Q&A

1:00 Off Label but On Target: Perspectives on and Examples of Drug Repurposing for Rare Diseases

Dr. Smith Heavner, Adjunct Assistant Professor, Sr Scientific Director, Real-World Evidence, Data Collaboration Center (DCC)

1:30 Symposium Q&A and Moderated Open Forum

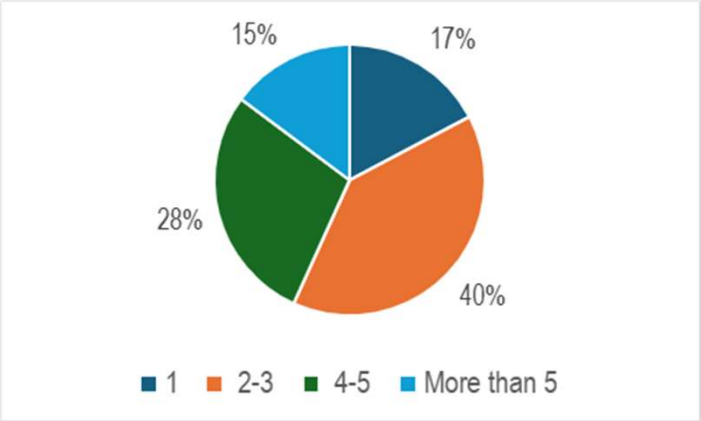
2:00 Adjourn

Appendix B. SC RDAC Needs Assessment Data

Key Themes

- Accessibility and Coverage
    - Travel and Accessibility: Covering travel expenses for patients who need to visit distant specialists or clinics.
    - Medication Affordability: Reducing the cost of medications and treatments to make them more affordable.
    - Insurance Coverage: Expanding health insurance coverage to include necessary treatments and medications.
- Education and Awareness
    - Provider Education: Educating healthcare providers about rare diseases, especially adult manifestations and specific conditions.
    - Patient Education: Providing more information and support to patients and their families about their conditions.
- Specialized Care
    - Access to Specialists: Ensuring patients have access to specialists who are knowledgeable about their rare diseases.
    - Coordination of Care: Improving communication and coordination between different healthcare providers and specialists.
- Support and Advocacy
    - Family Support: Offering assistance and resources to families of patients with rare diseases.
    - Advocacy and Resources: Providing access to resources, advocacy, and support groups to help patients navigate their healthcare journey.
- Research and Funding
    - Funding for Research: Increasing funding for research into rare diseases to develop better treatments and cures.
    - Compassionate Use Programs: Facilitating access to promising therapies that may not yet be FDA-approved.
- Healthcare System Improvements
    - Comprehensive Care Models: Establishing patient-centered care coordination models that ensure equitable access to treatments and support.
    - Telehealth and Satellite Clinics: Enhancing telehealth support and creating satellite clinics to reduce travel burdens for patients.
- Miscellaneous
    - Emergency Care: Ensuring appropriate care in emergency departments and holding providers accountable to guidelines.
    - Disability Support: Providing information and support for obtaining disability benefits and assistance.

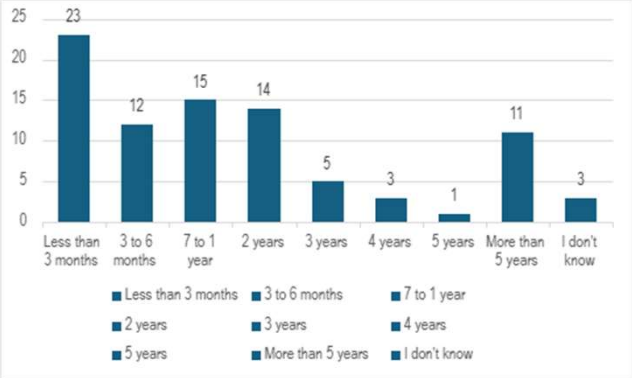
Approximately how many different physicians did the patient go to seeking diagnoses between the first signs or symptoms and the final diagnosis? (n=81)



43% of respondents reported that they had to go to more than 3 physicians before receiving a final diagnosis.

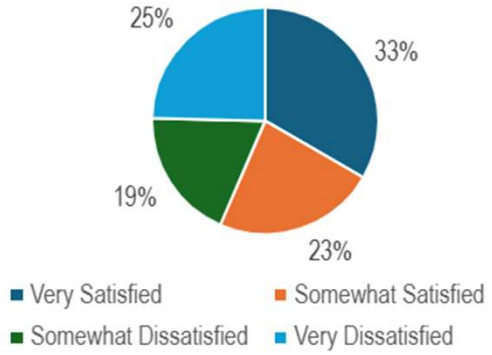
The average number of physicians respondents reported seeing is 4.

How long after first suspicion of a disease did it take to get an accurate diagnosis? (n=87)



39% of respondents reported that it took 2 or more years to receive an accurate diagnosis.

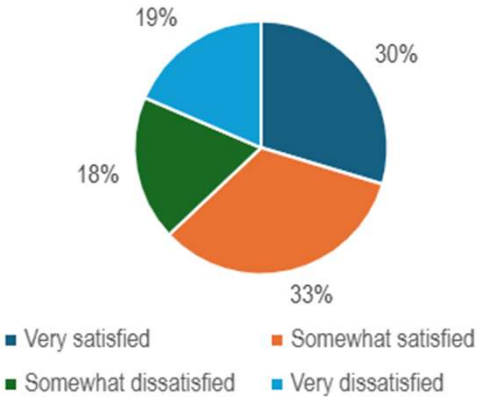
How satisfied was the patient with access to approved medications for the rare disease following diagnosis? (n=69)



What factors prevented the patient from being very satisfied with the access?

- Timeliness of Access (14)
- Cost (18)
- Travel (9)
- Other (22)
  - Few or no approved medications (8)
  - Lack of Insurance Coverage (5)
  - Side Effects (3)
  - Ineffectiveness (3)
  - Minimal data/limited recommendations from providers (3)
  - Medication not available locally
  - Excessive visits required

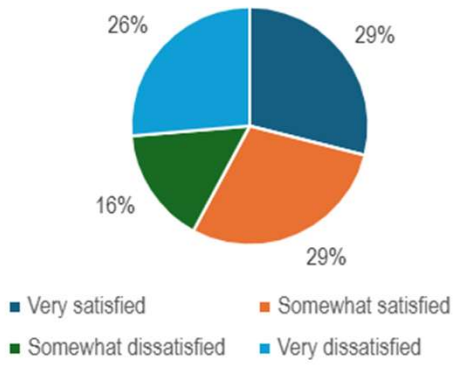
How satisfied was the patient with access to specialty care doctors/clinics following diagnosis? (n=81)



What factors prevented the patient from being very satisfied with the access?

- Timeliness of Access (21)
- Cost (17)
- Travel (29)
- Other (24)
  - Not enough doctors/no or few specialists; doctors not familiar with disease (1)
  - Felt misunderstood/judged at times (2)
  - Difficulty diagnosing (2)
  - Lack of coverage (2), not covered out of state (1)
  - Lack of continued follow up
  - Did not know about treatment centers
  - Support groups

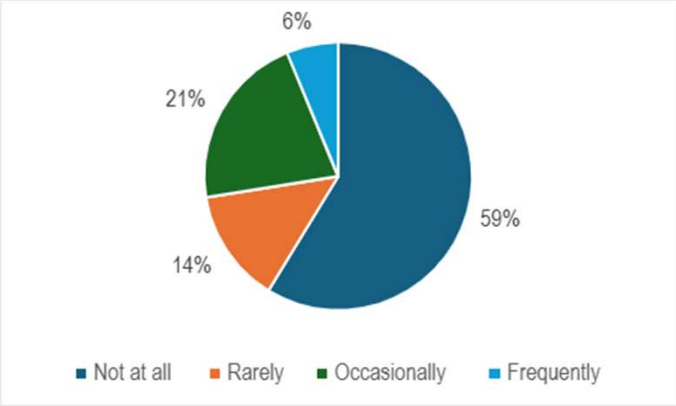
How satisfied was the patient with the access to further testing and procedures following diagnosis? (n=76)



What factors prevented the patient from being very satisfied with the access?

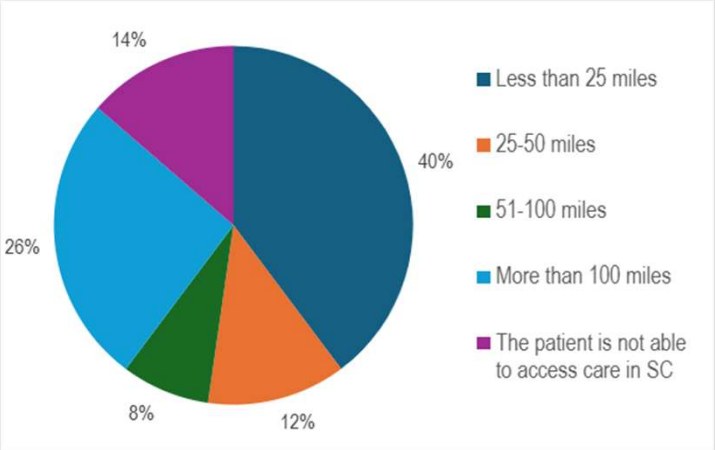
- Timeliness of Access (24)
- Cost (18)
- Travel (20)
- Other (24)
  - Information is/was not available or provided; doctors did not know about diagnosis (14)
  - Providers were dismissive (3)
  - Difficulty getting a referral (2)
  - Insurance denied (2)
  - Diagnosis of multiple rare diseases (1)
  - Information about alternatives (1)

How often, if at all, does the patient need to travel out-of-state for medical care related to the rare disease? (n=80)



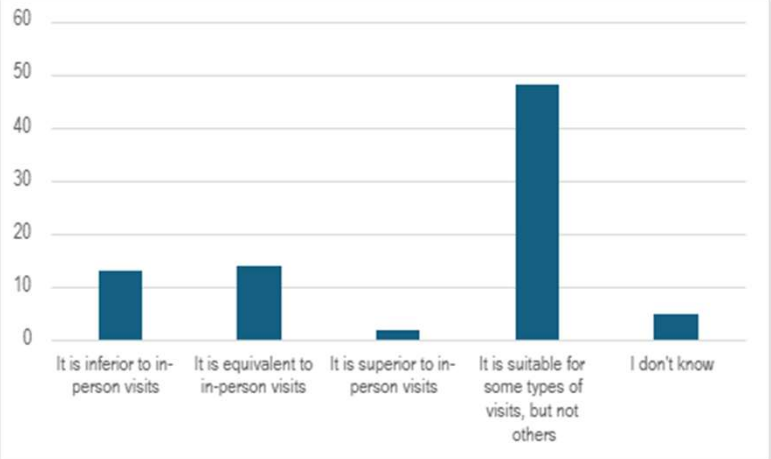
41% of respondents have to travel out-of-state for some or all medical care related to their rare disease.

How far does the patient need to travel within South Carolina for medical care related to your rare disease? (n=83)

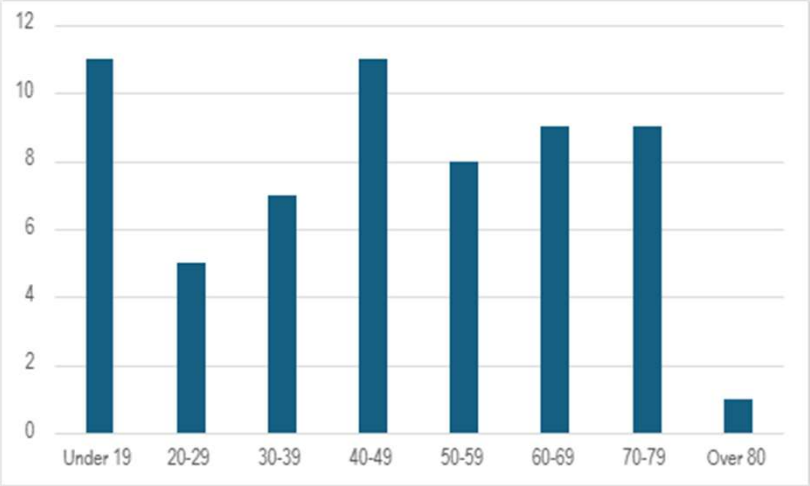


39% of respondents have to travel greater than 50 miles within South Carolina or have to travel out of state for medical care related to their rare disease.

What are the patient's general feelings towards telehealth? (n=82)

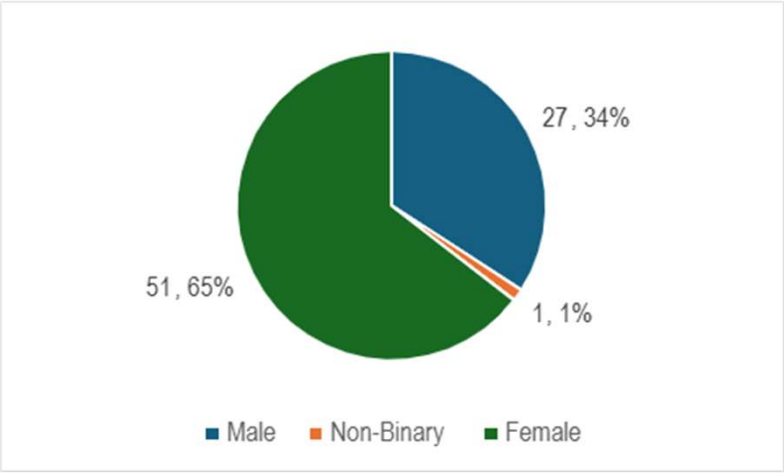


Demographics: Patient Age (n=61)



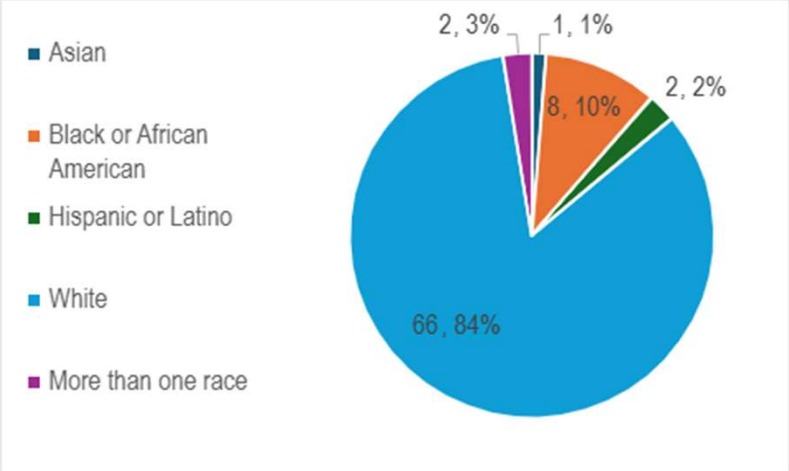
*Includes only respondents that completed the survey

Demographics: Patient Gender (n=79)



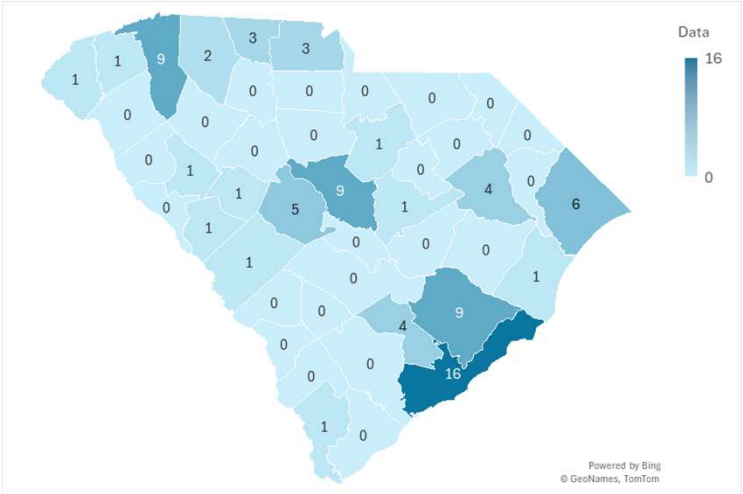
*Includes only respondents that completed the survey

Demographics: Patient Race (n=79)



*Includes only respondents that completed the survey

Demographics: Patient County (n=80)



*Includes all respondents that answered this question

Appendix C. Estimation of Rare Disease Prevalence in SC

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Professor
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The prevalence of rare diseases for the state of South Carolina (SC) is not known. This makes it difficult to estimate the services needed for these special populations. The following is a report for an exploratory study using two data sources for the year 2022 to develop and validate a method of estimation for two rare diseases with a relatively high prevalence: Polycystic Kidney (PolyK) disease and Sjogren's Syndrome (SJ). We then apply the approach developed to eight additional rare diseases.

Data Sources

The major source of data used was the SC All-Payer data (RFA data) base for the year 2022. The sources of these data include all bills, regardless of insurance type, for hospital admissions (HA), emergency department visits (ER) and outpatient surgeries (OP) that are provided by acute care hospitals and outpatients surgery centers in the state of South Carolina. These archival billing data are deidentified to preserve patient and provider confidentiality. The variables captured are based on data points required for filing the form UB04 medical facility form. Services provided to SC residents outside of the state are not included in the RFA data. Patient age, sex, race and ICD-10 codes for diagnoses and procedures and length of stay are included. This data source captures patients of all ages and by all insurers, as well as the uninsured population, who have a condition that involves hospital, ED or OP surgery. However, the rare disease may not always be coded as either the primary diagnosis or as one of 14 possible comorbid conditions. The RFA data does not capture visits in ambulatory settings, such as a medical office, Federally Qualified Health Center (FQHC) or outpatient clinic. To estimate the number of patients who used healthcare only in ambulatory settings, we used a second database for estimation of people only seen in an outpatient setting. These validation data were limited to include a 5% sample of patients with Medicare coverage. Thus, the validation database is specific for insured patients, contains only a sample of the population and has limited representation of age groups. We then adjusted the number patients with an RD diagnosis observed in the RFA data set to reflect the number of patients found using the Medicare 5% sample in the national Medicare 5% Limited Data set.

Data Analysis

We first extracted all records for SC patients with the rare disease (RD) conditions of interest from the RFA data. We included patients with ICD-10 code for the RD as a primary diagnosis or as one of 14 possible data fields for comorbid diagnosis. We constructed a demographic record for each

patient with an RD and summed the number of hospital admissions, ER and OP episodes recorded for each patient in the RFA data from 2022. We then extracted all records for patients with an RD diagnosis from the hospital admission, ED, and outpatient surgery care files for the MC data set for 2022 using the same approach as described for the RFA data set. We constructed a demographic record for each patient in the same manner as that used for the RFA data. We then calculated the number of individual patients (the unduplicated number of patients) that were included in the MC data set. This is the underlying population (rate denominator) that was the source of the RD events for each data set. We used the population denominator data and the MC RA population counts (numerator data) to adjust the RFA RA patient numbers to reflect the number expected if patients with only office visits who would have been included in the RFA data if these data contained CMS forms events. We report both the actual observed RFA RD numbers and the estimated total that capture patents who only received office-based care in 2022.

Results of Data Analyses

The data analysis for the RFA data yielded the number of patients with each RD and the number of HA, ER and OP events recorded for each person for groups defined by age and insurance. However, these summary data excluded patients with the RD who did not use any hospital, ED or OP services in 2022. The MC data set yielded data from subgroups of patients by age and race with Medicare insurance, but these data contained only a 5% random sample of Medicare beneficiaries which is a small fraction of the SC population. Thus, the RFA data set was missing patients with the RD diagnosis coded who only had medical office visits. This is because offices use a different billing form (CMS 1500) from institutions who use the UB04 form for payment. The use of the MC data in addition to the RFA data enables us to estimate the number of office-visits-only RD patients missing from the RFA data set. The detailed estimates for Polycystic Kidney disease are shown in Table 1 below. The estimates for 10 rare diseases with and without the adjustments for missing office visits are provided in Table 2.

Table 1: Number of SC Medicare Beneficiaries in 2022 with a Diagnosis of Polycystic Kidney Disease as Estimated from RFA All-Payer Data and the Medicare 5% Data Sets.

Data Set	Hospital Cases N (%)	ER, OP and Office Cases N (%)	**Total Observed**	Population Size	**Total Estimated**
Medicare 5% sample for SC	120 (32%)	260 (68%)	**380**	1,223,960	NA
RFA Data	115 (32%)	244 (68%)	**359**	1,131,048	**388**
Population Adjustment Factor				1.082147	

The data in Table 1 shows that the Medicare 5% data set drew from a large population base, and that the number of Medicare patients captured by the SC RFA data set was only slightly smaller. Further, the distribution of patients with a PolyK diagnosis found in the Medicare 5% sample data was identical for the percentage identified from hospital admissions records and the percentage found in OP, ER data (32% and 68% respectively). Thus, the RFA estimate for patients with PolyK diagnosis needed only adjustment by the population size factor (multiplied by a factor of 1.082147) representing the differences in the underlying denominator population. Thus, the estimated number of Medicare patients with PolyK diagnosis in SC in 2022 is 388.

Table 2: Results for 10 Rare Disease Estimated Using the SC All-Payer Data Set for 2022 with Adjustment for Missing Cases with Only Medical Office Visits in the Year.

Rare Disease Name	Observed Cases	Estimated Cases	Age Median (range)	Percent Female	Percent White	Percent Black	Percent Other
Polycystic Kidney Disease	731	928	53 (1-85+)	54%	56%	38%	6%
Sjogren's Syndrome	4,452	6,811	60 (1-85+)	93%	74%	22%	4%
Scleroderma	1,888	3,055	59 (4-85+)	39%	68%	29%	3%
Polycythemia Vera	1,277	2,363	63 (3-85+)	39%	86%	12%	2%
α 1 Antitrypsin Deficiency	660	900	63 (2-85+)	57%	94%	4%	2%
Duchenne Muscular Dystrophy	127	209	18 (1-70)	15%	72%	16%	2%
Von Willebrand's Disease	997	1,274	38 (1-85+)	78%	82%	13%	5%
Cystic Fibrosis	793	793	22 (1-85+)	57%	81%	14%	5%
Amyotrophic Lateral Sclerosis	671	1,063	62 (19-85+)	44%	76%	20%	4%
Guillain Barre Syndrome	1,477	1,618	57 (3-85+)	52%	76%	20%	4%

The results in Table 2 above show the number of unduplicated patients identified in the SC All-Payer data sets for 2022 for each of the ten conditions examined in this pilot study. Colum two show the estimated number of unduplicated cases for 2022 calculated from the number of additional patients identified in the Medicare or Commercial Insurance data samples used for validating the numbers observed in the RFA data set. The third column shows the median age and age range

observed in the RFA data. The last four columns in the table show the percentage of patients who were female, and the racial distribution observed in the RFA data.

We successfully developed and validated an approach to estimate the prevalence of rare diseases in SC. The best, most comprehensive data set turned out to be the RFA All-Payer data. The best data set for validation was the Medicare LDS 5% sample so this was used for all diseases for which the Medicare data did not show great age limitations. For younger patients we used the MarketScan Commercial data to capture patients who did not have an ED, OP surgery or hospital admission during the year. This approach was needed to capture individuals who received all their medical care in a physician's office and therefore had all bills submitted on the CMS 1500 form and no available records filed on the UB04 form. This approach was checked using a MarketScan Medicare database, and found to capture a prevalence that varied by less than 10 individuals for each RD.

In summary, we developed and validated an approach to estimating RD prevalence for SC using the RFA All-Payor data. We were able to estimate the number of patients who were not captured in the RFA data. The proportion of RD patients missed by the RFA data varied by disease. However, the statistical program used (SAS version 9.4) for extraction worked across all conditions once we had identified the key ICD-10 diagnosis codes needed to extract the data for each RD condition from the RFA data and from the Medicaid or Commercial data. A drawback of this approach is that this requires data extractions from up to six different data tables for each condition, and each data run takes between 30 minutes and 4 hours. This programming and computer time use could be vastly reduced if we repeated the analysis for the 10 current conditions on data from 2023 or a later year. If this analysis showed the outpatient visit adjustment factor for each condition was stable over time, which we expect it will be. Once this is done, then we can reduce the programming and computing time by at least 50%, as well as make updates as RFA data become available.

We suggest that we repeat the present analysis on 2023 data to derive outpatient (part B) adjustment factors from the other data sources. Once that is done, the data requirements and programming time will decrease tremendously. Thus, it will become feasible to estimate the next 100 RD conditions rapidly and economically.

In summary, we have successfully developed and tested an approach to identifying the prevalence of RD in SC using the state's available data. We have validated computer code that only needs ICD-10 codes replaced in the array finder statements and in the prevalence calculation steps for the code to be applied to other RD conditions. This has been a challenging but rewarding project and we hope to continue the work on more RD conditions in the future.

Appendix D. SC RDAC Quarterly Meeting Minutes

Rare Diseases Advisory Council

July 26, 2024

Meeting Minutes

1. Welcome
2. Introductions
3. Newborn Screening Protocols presentation by HCU Network America Communications Manager Liz Carter
    a. **Goal of HCU NA is to get the methionine level at birth cutoffs lowered from 65 to reduce the likelihood of missing a diagnosis of classical HCU at birth.**
    b. **HCU NA has an upcoming meeting with the SC NBS lab meeting to discuss recommended newborn screening recommendations and new assays discovered by the CDC for improving HCU detection (Dr. Chapaigne was invited to join).**
    c. **Neena Champaigne discussed her time and efforts on the SC NBS lab advisory council.**
        i. **Explained the importance of getting on having concrete numbers reflecting at birth methionine levels and delayed diagnosis.**
        ii. **Explained potential risks of lowering the methionine levels too much (ex. Typically raised levels in preemie population), but also suggested that there were potential tiered testing solutions that should be explored.**
        iii. **Expressed interest in future inclusion in HCU NA's interactions and efforts with SC PHL**
    d. **Council follow-up items: Determining what action items requested by Liz that the RDAC can/wants to pursue:**
        i. ***SCRDAC Considerations***
            1. Would you consider including a question re: missed NBS cases in SC on your Rare Disease Needs Assessment?
            2. Would you consider putting an excerpt on your website or in a newsletter from HCUNA re: this issue?
            3. Would you consider communicating a recommendation on the need to review NBS protocols for HCU to your state NBS program?
        ii. **Patrick Flume suggested that we invite someone from NBS lab to join and SC RDAC meeting to explain their process and decision-making workflow**

4. SC RDAC Charter
    a. **The council supports Dr. Flume's efforts to have the terms of council members changed.**
    b. **Ginie Daguise suggested having both 2 and 3-year terms so that there is not a loss of institutional memory**

c. **Karen Kemper suggested that after the initial term, the council could renew members for up to an additional set number of terms. She explained that this has worked well for other advisory groups of which she has been a part.**
d. **Jonathan Hawayek said that some councils have an initial term and then, with vote from the council, a member's term could be extended to a different amount of time.**
e. **Jonathan Hawayek suggests we be cautious in bringing on too many people to the council, as it could become too difficult to manage. He suggests bringing people on an ad hoc basis or the use of subcommittees to leverage the expertise of certain individuals depending on council initiatives.**
f. **Some states do not allow for the RDACs to lobby/testify for legislation or offer letters of support for initiatives. When developing the SC RDAC charter, we should confirm that we are not limited in that way.**
g. **Dr. Flume suggested we consider adding a new section to the charter, under the council's responsibilities, outlining the ways in which the RDAC might offer recommendations to the state.**
h. **Council follow-up items:**
    i. **Dr. Flume to look into getting permission to edit council terms of service**
    ii. **Dr. Flume to confirm that we aren't limited in ways we can lobby or offer support for initiatives.**

5. Housekeeping and Additional Topics
    a. Recommendation for Rare Disease Organization representation
    b. SC-RDAC Progress Report was submitted to the SC Governor
    c. SC Rare Disease website updates (scraredisease.org)
    d. **The council asked for an update on the needs assessment.**
    e. **Council follow-up items:**
        i. **Jonathan Hawayek to contact NORD for potential recommendations on who to fill our RDO council spot.**
        ii. **Jonathan Hawayek will provide codes for prevalence and incidence data available for 1 through 500 to Ginie Daguise to take them to get epi data from the state.**
        iii. **Jonathan Hawayek will find contact information for Minnesota RDAC needs assessment so we can invite them to meet with us to discuss their findings and the development and distribution of the survey.**
6. Adjourn

Membership Attendance:

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
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) Neena Champaigne, MD (Pediatric Genetics) Chip Norris, PhD (MUSC, connective tissue)	A P A
Patient (2)	Anthony Hamilton (familial TTR amyloid) Karen Kemper, PhD (scleroderma)	A P
Rare Disease Organization	N/A	
Caregiver of person with rare disease	Jason Smith (hypoplastic left heart syndrome)	A
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	P

Non Member Attendance: Liz Carter (HCU Network America), Danae Bartke (HCU Network America), Holly Murphy, Kevin Mann (Novo Nordisk), Sara Stolfus (Artia Solutions), Samantha Darwak, Tara Tsagouris (Jazz Pharma), Kemi Olabisi (Acadia Pharmaceuticals)

Rare Diseases Advisory Council

October 25, 2024

Meeting agenda:

1. Welcome
2. Introductions
    a. Halley Harris was introduced as the new Administrative Coordinator

3. Newborn screening process presentation by Tanya Spells, Director of Newborn Screening Follow-up for S.C. Department of Public Health
    a. Slides shared with council members
    b. RDAC will post the link to SC newborn screening resources on the RDAC webpage

4. Updates from NORD's RDAC Member Meeting and the Rare Diseases and Orphan Products Breakthrough Summit (Dr. Neena Champaigne and Tara Pittman, attendees)
    a. Key Take Aways from Members Only Meeting
        i. The theme for this year's meeting was Equitable Access to Innovation
        ii. 29 States now have RDACs
            1. Funding is an ongoing issue with initiatives
        iii. Minnesota working on PA reform with Medicaid to eliminate the burden of refiling every 6-12 months for patients and providers
        iv. Prescription Drug Affordability
            1. Cost of orphan drugs
    b. New resource portal for RDAC members and administrators (includes slides from the 2024 member meeting): https://rdac.rarediseases.org.

5. SC RDAC Charter
    a. The adding and removal of members
        i. When/about what do we vote on? Do we need to wait for in person meetings? How do we define "inactivity" or ground rules for membership?

    b. Dr. Flume requested volunteers to assist with finalizing charter.

6. Housekeeping and Additional Topics
    a. Committee member progress on follow-up items from last meeting (see July meeting notes)
        i. Jonathan Hawayek spoke with Carolyn Sheridan from NORD about helping us find a replacement member to represent RDOs. She will get back to us with potential suggestions.
        ii. Jonathan Hawayek received codes for prevalence and incidence data available for 1 through 500 and requested for them to be more user friendly. He is expecting them to be ready in the next few weeks.
        iii. Jonathan Hawayek was given Erica Barnes as administrative contact for Minnesota RDAC.

b. Ideas on how to handle requests for information from the community
    i. Looking for referrals/access to information, seeking patient connections for media opportunities, etc.

c. Administrative Updates:
    i. Needs assessment is being advertised through the Post & Courier (in print and digitally) across the state through the month of October.
        1. 13 ads across the state of SC
        2. 200,000 digital banners
        3. 10 completers of the survey, 98 people, 57 fully completers
        4. Vicki Young suggested sharing ad from Post & Courier and digital banner to advertise at Community Health Center sites, clinical sites, and newsletters
        5. Sonya Rigsby suggested partnering with Patient Advocacy groups to share in newsletters. Follow up on obtaining contacts from her.
    ii. Segra Park in Columbia has been reserved, once again, for next year's SC Rare Disease Symposium to be held on February 28th (Rare Disease Day)

d. Follow Up Items
    i. Tara will be reaching out to Erica Barnes at University of Minnesota to see if someone from their RDAC is available to speak about the promotion of their needs assessment survey at our January meeting
    ii. Ginie Daguise to get prevalence and incidence data from Jonathan Hawayek in next few weeks and take them to get epi data from the state (waiting for the full 500)
    iii. Vicki Young and Patrick Flume to meet and discuss other state data available for review
    iv. RDAC admin to organize specialized meetings to be held before the end of the year to finalize the charter and discuss the symposium. (Volunteers: Kate Wink, Karen Kemper, Jonathan Hawayek, and Ginie Daguise)

7. Adjourn

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)	P
Medical University of South Carolina	Dan Judge, MD (MUSC, cardiology)	P
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	P
South Carolina Hospital Association	Kate Wink (Santee Cooper)	P

South Carolina Primary Healthcare Association	Vicki Young (SCPHCA)	P
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)	A P A
Patient (2)	Karen Kemper, PhD (scleroderma) 2nd patient N/A	 P
Rare Disease Organization	N/A	
Caregiver of person with rare disease	Jason Smith (hypoplastic left heart syndrome)	A
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	P
Program Coordinator	Halley Harris	P

Non Member Attendance: Tanya Spells, Sonya Rigsby, Michelle Meyer, Camerun Washington, Vyndra Smith, Samantha Darwak,

Rare Diseases Advisory Council January Quarterly Meeting
Meeting Minutes
January 10th, 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 28th @ Segra Park (Columbia, SC)**
    a. Draft Agenda

Time	Title/topic	Speaker
9:30	Check In/ Registration	
10:00	Welcome/opening remarks	Dr. Patrick Flume
10:10	State of the state • Show data (Ginie Daguise) and/ or report card	*Jonathan Hawayek to reach out to Carolyn Sheridan from NORD for availability*
10:30	Changing policy in South Carolina • Approaches to Rare Disease in Legislation, Challenges in Legislation	Ken Bingham or Emily Bingham *Follow up on availability from Jonathan Hawayek*
11:00	Discovery/translational science • Zebrafish	Greenwood Genetics Mike Lyons or Heather Rich *Follow up on availability from Steve Skinner*
11:45	Networking Lunch	
12:00	Personal story (led to doing science)	Tara O'Sullivan *Follow up on availability from Neena Champaigne*
12:30	Personal story: Participation in clinical trial	Cara O'neal *Follow up on availability from Neena Champaigne*
1:00	Q&A for Personal Stories	
1:30	Advocacy development • FDA/critical path institute	Smith Hevner *Follow up on availability from Karen Kemper*
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) Neena Champaigne, MD (Pediatric Genetics) Chip Norris, PhD (MUSC, connective tissue)	P P
Patient (2)	Karen Kemper, PhD (scleroderma) Bridget Downing	P 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 Tara Pittman	P 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.

Rare Diseases Advisory Council Quarterly Meeting
April 11th, 2025
9:00 AM-10:00 AM

Meeting agenda:

- Welcome & Introductions
  - Yvonne Donald, M.A., CSCEC (James R. Clark Memorial Sickle Cell Foundation)
  - Cara O'Neill, MD, FAAP (Cure Sanfilippo Foundation) | Not Present
  - Kimberly Brown (RDAC coordination)

- Annual Report

  The RDAC annual report is due on June 30, 2025. The group discussed the SC Report Card from NORD, evaluating the state of South Carolina in comparison to surrounding states, and deciding which policy recommendations to prioritize in the report. The categories discussed include:
  - Medicaid Financial Eligibility
  - Medical Nutrition
    - The group discussed potential ways to impact medical nutrition outside of recommendations to the legislature, with specific consideration for what Florida has implemented.
    - Next steps:
      - Reach out to NORD to better understand what Florida has implemented and share with Dr. Wessinger.
      - Was able to find the medical coverage guideline for enteral formulas on the Florida Blue website which references the state mandate for coverage in this area.
  - Newborn Screening
    - The group discussed ongoing work and expected traction in the area of newborn screening. The group discussed monitoring an ongoing pilot in Florida which offers free genetic testing for newborns.
    - Next Steps:
      - Dr. Champaigne has reached out to the NBS Advisory Committee members to see if we can have and RDAC member join the committee. Tara and Dr. Flume have been cc'd on the email and will coordinate next steps after receiving a reply.
  - Prescription Drug Out-of-Pocket Costs
    - The group suggested that this be a prioritized recommendation in the annual report.
  - Protecting Patients in State Medicaid Programs
  - Protecting Patients in State Regulated Insurance
    - Next steps:
      - Here is a helpful description on short term limited duration insurance (STLDI) plans. In summary, they are short term plans that

are not regulated by the ACA or other federal insurance rules, meant to fill a gap in coverage (not like COBRA, which allows for a continuation of an existing employer insurance coverage). They are not comprehensive and are mostly unregulated at the federal level because they are not considered individual health insurance like ACA compliant plans sold through Marketplace. There was a finalized federal rule that came out in March of 2024 limits new STLDI plans, sold or issued on or after September 1, 2024 to three-month terms, and caps total durations (ie. renewals) at no more than 4 months. States can have stricter rules on STLDI plans than the federal rule, but not more lenient. SC had stricter regulations than federal rules from 2018- Aug 2024 but now federal rules are tighter with the changes that took place in Sept 2024.

- You can find a summary of the recent federal rule change, the reasons for the change, and where certain states fall in their restrictions of STLDI plans here.

- NORD's suggestions for improving a grade in this area is to more strictly regulate short-term plans (beyond federal rules). Examples of policy options for doing so include: banning short term plans, further limiting plan duration, limiting "stacking" of different STLDI plans, implementing preexisting condition protections, limiting premium rating factors, limiting who can enroll, prohibiting rescissions..
    - Here is the link to NORD's webpage describing STLDI and their rubric for grading

- Rare Disease Advisory Council
- Step Therapy (Fail First)
    - The group discussed benefits and challenges related to step therapy and step therapy policy.
    - Next steps:
        - Reach out to NORD to see if they have data/evidence to demonstrate the hindrance of response time.
- Telehealth
    - The group discussed the benefits and challenges related to telehealth.
    - Next steps:
        - The group suggested that this be a prioritized recommendation in the annual report, pending what Jon Hawayek finds out regarding context of previous legislative discussions related to the Interstate Medical Licensure Compact (IMLC).

- Charter & Succession Planning
    - Did not get to discuss this item. The charter was approved by council via electronic vote and RDAC will now adhere to those guidelines going forward.

- 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)	P
Department of Health and Environmental Control	Ginie Daguise (SCDHEC)	P
Department of Health and Human Services	Kevin Wessinger (SCDHHS)	P
Medical University of South Carolina	Dan Judge, MD (MUSC, cardiology)	A
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)	A
South Carolina Primary Healthcare Association	Vicki Young (SCPHCA)	P
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)	P
Caregiver of person with rare disease	Cara O'Neill, MD, FAAP (Cure Sanfilippo Foundation)	A
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	P

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.