South Carolina Rare Disease Advisory Council

Rare Diseases in South Carolina

2025-2026 Annual Report

MUSC
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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 26, 2026

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 Diseases Advisory Council, it is my pleasure to submit the third annual legislative report as outlined in Act 94 of 2021, the FY 2021-2022 Appropriation Act. 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 established by Act 94 of 2021, the FY 2021-2022 Appropriation Act; there are currently 32 states that have enacted legislation to establish an RDAC.[1] The South Carolina General Assembly, through Proviso 117.144 (GP: Rare Disease Advisory Council) of the FY 2025-2026 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. The council's mission, membership, and official responsibilities are provided later in this report.

1b. Summary of council initiatives

Since the last progress report submitted in June 2025, the council has tackled several initiatives as outlined in its official responsibilities. Main accomplishments of the council during the 2025-2026 year are highlighted below and will be discussed in greater length throughout the report:

- Held quarterly meetings open to the public to encourage statewide collaboration on, and support of, council initiatives;
- Hosted the third annual Rare Disease Symposium on February 27, coinciding with National Rare Disease Day (February 28), to promote research and spread awareness of and advocacy for the state's rare disease community;
- Presented at state and national levels, and collaborated with other state RDACs, to increase visibility and impact, share lessons learned, and promote the work of SC RDAC;
- Bolstered the official SC RDAC website (rarediseasesc.org) to optimize awareness of state and national resources;
- Developed a process to triage outreach from the public to better meet the needs of the rare disease community and foster engagement; and,
- Broadened pilot study efforts 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.

SC RDAC continues to monitor updates to the Rare Disease State Report Card developed by the National Organization for Rare Disorders (NORD) to assess progress on key rare disease–related issues in South Carolina and compare the state's landscape with that of peer states.[2] In addition, the Council has developed an internal process to track proposed legislation that may affect the rare disease community. Monitoring these bills helps the SC RDAC identify emerging areas of need and interest across stakeholders, which in turn informs the Council's prioritization of initiatives related to resource development and dissemination, educational opportunities, and data collection efforts.

The minutes from the quarterly meetings of the SC RDAC and the expense report follow in this report.

[1] National Organization for Rare Disorders, https://rarediseases.org/policy-issues/rare-disease-advisory-councils/
[2] NORD State Report Card, https://rarediseases.org/driving-policy/nord-state-report-card/

2. SC RDAC Mission and Membership

2a. Background

Under the U.S. Orphan Drug Act, a rare disease is defined as a condition affecting fewer than 200,000 people nationwide.[3] Although rare individually, these conditions collectively affect more than 30 million Americans across over 10,000 diseases.[4] Many rare diseases are serious, progressive, or life-threatening, and most still lack an FDA-approved treatment, leaving patients with significant unmet medical needs.[5]

The impact of rare diseases extends beyond health outcomes. Individuals and families often face delayed diagnoses, limited access to specialized care, and reduced quality of life—challenges that translate into a disproportionately high economic burden on society.

A 2022 analysis supported by IQVIA, global provider of advanced analysis to develop, test, and maintain new treatments, and conducted by Chiesi Global Rare Diseases found that, on a per-patient, per-year basis, the economic burden of rare diseases is approximately 10 times higher than that of mass-market diseases, even before accounting for broader social costs such as diminished health-related quality of life. The study also showed that available treatments can reduce portions of this excess burden, underscoring the value of continued investment in rare disease therapies, diagnostics, and screening programs.[6]

Together, these findings highlight the importance of informed policy responses. The SC RDAC enables government leaders to better understand the real-world impact of rare diseases at both the individual and state levels, supporting data-driven legislative decisions that improve outcomes for people living with rare conditions.

Rare Disease Quick Facts

Americans Affected

Rare Diseases are individually uncommon but collectively widespread.

Delayed Diagnosis

Patients often wait years for an accurate diagnosis due to limited awareness and testing.

Few Treatment Options

Over 90% of rare diseases lack FDA-approved therapies.

Fragmented Care

Patients face challenges navigating specialists, research, and support systems.

[3] US Food and Drug Administration, https://www.fda.gov/patients/rare-diseases-fda
[4] Ibid.
[5] Ibid.
[6] Chiesi Global Rare Diseases, https://chiesirarediseases.com/assets/pdf/chiesiglobalrarediseases.whitepaper-feb.-2022_production-proof.pdf

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 sixteen members and is 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 Public Health (SC DPH), the Director of the Department of Health and Human Services (DHHS), the Executive Director of the South Carolina Hospital Association (SCHA), the Chief Executive Officer of the South Carolina Primary Healthcare Association (SCPHA), the Executive Director of the State Public Benefit Authority (SPBA), and the President and CEO of Greenwood Genetic Center.

As the Council enters the new fiscal year, leadership will transition from Dr. Patrick Flume to Dr. Neena Champaigne as Chair.

Table 1. SC RDAC Member Composition

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
Greenwood Genetic Center	Steve Skinner, MD
Prisma Health - University of South Carolina	Divya Ahuja, MD, MRCP (infectious disease)
South Carolina Hospital Association	Kate Wink, MPA (Santee Cooper)
South Carolina Primary Healthcare Association	Vicki Young, PhD
State Health Plan	Tripp Jennings, MD (BCBS South Carolina)
Biopharma Industry	Jonathan Hawayek (Genentech)
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, MA, CSCEC (James R. Clark Memorial Sickle Cell Foundation)
Caregiver of person with rare disease	Cara O'Neill, MD, FAAP (Cure Sanfilippo Foundation)

2d. Responsibilities of the Advisory Council

Stakeholder Engagement

Gather input from stakeholders across South Carolina, including patients and caregivers affected by rare diseases, to identify needs and challenges.

Expert Consultation

Consult with rare-disease experts to develop recommendations that improve access to specialists, diagnostics, treatment, and comprehensive, affordable health care.

Research & Policy Priorities

Research and identify priorities related to treatments and services provided to persons with rare diseases and develop recommendations.

Education & Awareness

Publish a list of existing, publicly accessible resources on research, diagnosis, treatment, and education. Identify and share educational resources to support prevention, recognition, and optimized treatment of rare diseases.

Health Equity

Identify best practices to reduce health disparities and achieve health equity in the research, diagnosis, and treatment of rare diseases.

Transparency & Reporting

Maintain a public website with annual reports, meeting notices and minutes, and resources. Submit an annual report to the Governor and General Assembly reporting SC RDAC activities, progress, and recommendations.

3. Projects and Achievements

3a. Operations, Activities, and Outreach

During the past year, SC RDAC held quarterly virtual meetings (July 2025, October 2025, January 2026, April 2026). These meetings were open to the public, with Microsoft Teams links available on the SC RDAC website.

Dr. Champaigne presenting at the NORD Rare Action on the Road event in Columbia, SC.

Expanded Public Outreach

Dr. Neena Champaigne presented at the NORD Rare Action on the Road event in October 2025, which brought together individuals affected by rare diseases, caregivers, clinicians, and organizational partners. The event featured expert speakers and interactive sessions designed to share information, build skills, and foster connections within the rare disease community. At the event, SC RDAC shared its annual report.

SC RDAC also engaged with stakeholders by tabling at professional and community events, including the South Carolina Clinical and Translational Research (SCTR) Institute Scientific Retreat, which convened researchers, clinicians, trainees, patients, and industry partners to focus on cellular and gene therapy. In addition, the Council participated in the MUSC Center for Healthy Aging Senior Expo, a free community event serving seniors, caregivers, and families across the Lowcountry, which featured health screenings, educational resources, and exhibitors and drew approximately 800 attendees. Furthermore, SC RDAC hosted its Third Annual Rare Disease Symposium, which is discussed in more detail in Section 3c.

Strengthened National Collaboration

Council members Dr. Champaigne and Bridget Downing, along with administrators Tara Pittman and Kimberly Brown, represented SC at NORD's 3rd Annual RDAC Leadership Meeting in Washington, DC. The meeting included representatives from 25 state RDACs and featured sessions on state budgets, FDA updates, Medicaid integration, operational best practices, and sustainability strategies. Following this meeting, the group attended NORD's two-day Rare Disease and Orphan Products Breakthrough Summit; Dr. Champaigne and Ms. Downing's associated costs were covered by NORD.

In addition, Kimberly Brown presented a poster at the National Fall Clinical and Translational Science Award (CTSA) Program Annual Meeting titled *Embedding a State Rare Disease Advisory Council Within a CTSA Hub to Advance Community-Driven Translational Research*. The presentation highlighted how SC RDAC's model supports stakeholder engagement, enhances research infrastructure, and informs community-driven research priorities.

Group photo of RDAC representatives at NORD's 3rd Annual RDAC Leadership Meeting in Washington, DC.

3b. Enhancing Public Information and Responsiveness to Community Inquiries

To improve transparency and access to information, SC RDAC enhanced its online presence and strengthened its approach to responding to public inquiries. A new policy-focused webpage was developed to provide accessible, up-to-date information on rare disease–related legislation and policy activity relevant to South Carolina.[7] This page includes tracking of specific state bills (Table 2) with the potential to affect the rare disease community and serves as a centralized resource for patients, caregivers, providers, and stakeholders seeking information on policy developments.

Table 2. SC Bills of Potential Relevance to the Rare Disease Community (126th General Assembly, 2025-2026)

Bill	Summary
H. 3934	Cost Sharing, Copay Accumulators
S. 330	Cost Sharing, Copay Accumulators
S. 531	Healthcare Services, Step Therapy
H. 4335	Telehealth and Telemedicine Providers
S. 377	Telehealth and Telemedicine Providers
S. 717	Mother and Newborn Ombudsman
S. 749	Prescription Medicine Study Committee
H. 4790	Pharmacy Benefits Managers – Max Allowable Cost Lists
H. 4791	Drug Acquisition Cost Surveys and Reimbursements
H. 4794	Pharmacy Benefits Managers – Trade Practices
H. 4795	Pharmacy Benefits Managers – Drug Modifications

[7] SC RDAC, https://rarediseasesc.org/policy

Recognizing the growing volume and complexity of inquiries submitted through the RDAC “Contact Us” web form, SC RDAC also implemented a process to systematically track the types of requests received. This effort enabled the RDAC to better understand community needs and to create a more formalized approach to responding and connecting individuals with available resources, where possible. In parallel, RDAC leadership engaged with other state RDACs to share best practices related to inquiry management, public-facing communication, and community support.

The inquiries received underscore persistent gaps in care, information, and coordination for individuals affected by rare diseases. Requests commonly relate to the following topics:

- **Disease-Specific Information and Resources**
  - Requests for condition-specific education, awareness materials, and advocacy resources
- **Care Navigation and Access Challenges**
  - Assistance navigating complex health systems and identifying appropriate care
  - Help locating specialty providers within South Carolina
  - Concerns related to gaps in care, delayed treatment, or limited access to services
- **Financial, Disability, and Patient Rights Concerns**
  - Requests for information on financial assistance programs
  - Questions regarding disability benefits and approval processes
  - Reports of perceived patient neglect, discrimination, or barriers to care
- **Research and Clinical Referral Opportunities**
  - Interest in research participation
  - Provider requests for referral pathways to connect patients with research opportunities

Collectively, these requests demonstrate a clear need for coordinated, accessible support for South Carolinians affected by rare diseases. By improving online resources, tracking inquiries, and refining response processes, the RDAC is better positioned to identify community needs, inform future priorities, and connect individuals to appropriate information, partners, and opportunities.

3c. Third Annual South Carolina Rare Disease Symposium

The council held its 3rd annual Rare Disease Symposium on February 27, 2026, coinciding with the National Rare Disease Day (February 28). 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 130 attendees (65 in-person, 65 virtual). 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.

The symposium focused on a central theme: the transition from pediatric to adult care for individuals with rare diseases, an increasingly important issue as life expectancy improves. Discussions

emphasized both the progress made and the substantial challenges that remain. The agenda (Appendix A) covered several key areas, including:

- An overview of rare disease prevalence in South Carolina
- Health economics and ethical decision-making in rare disease care
- Non-medical aspects of transition, such as support systems, training, and care coordination
- Resources, navigation tools, and family-centered approaches to transition

A panel session also highlighted real-world experiences with transition including a rare disease patient and parent describing the process of supporting their child through the transition to adulthood, and a clinician offering insights on best practices and systemic needs within transition pathways.

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

- "The variety of experience available that ranges from providers, to support sectors (economics, statistics, etc.), to patients & caregivers who were able to be very open and vulnerable about things was amazing."
- "The most valuable aspect of the symposium was the emphasis on longitudinal continuity in rare disease care, particularly the integration of real-world systems and transitional frameworks across the lifespan."

Dr. Patrick Flume speaking at the Third Annual South Carolina Rare Disease Symposium.

An article titled "*Stripes, Science, and Solidarity at the 2026 Rare Disease Symposium*" was featured in MUSC Catalyst News and is also available on the SC RDAC homepage. The article gained national visibility and was picked up by additional outlets, including the National Center for Advancing Translational Sciences (NCATS) CTSA News.

SC RDAC intends to hold the 2027 Rare Disease Symposium at Riverbanks Zoo & Garden (SSA Group, LLC) in Columbia, SC.

3d. 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. This work lays the foundation for a scalable, cost-effective method to

support evidence-based policy decisions. This section provides a brief overview of the methods, analysis, and findings; the full report is included in the Appendix (Appendix B).

This report presents estimates of rare disease (RD) prevalence in South Carolina (SC) for 38 selected diagnoses identified among the most prevalent conditions listed in the Artia Solutions Rare Disease Summary Inventory and corresponding ICD-10 codes, provided by Genentech.

Data from the South Carolina All-Payer Dataset (2018 through mid-2024) were used to estimate prevalence based on hospital admissions and emergency department (ED) visits. These estimates were adjusted to account for individuals receiving care exclusively in outpatient settings, using data from Medicare and commercial insurance sources. Healthcare utilization metrics—including total hospital admissions, hospital days, and ED visits—were calculated, and total healthcare system costs were derived from recorded charges using a 27% cost-to-charge ratio. Annual per-patient costs were estimated from a payer perspective, with Medicare data informing estimates for older populations and commercial insurance data used for younger populations.

The 38 RD conditions examined correspond to approximately 29,300 unique South Carolina residents. County-level prevalence rates ranged from 0.04 to 1.65 RD patients per 1,000 population. Higher rates were observed in counties with greater availability of medical resources, suggesting potential underdiagnosis in areas with lower provider density.

In 2023, individuals with these RD conditions accounted for an estimated 13,000 hospital admissions, 76,500 hospital days, and 25,000 ED visits. The total cost of care was approximately $490 million in 2024. Mean annual per-person costs varied widely across conditions, ranging from approximately $8,000 to more than $200,000. These findings highlight the substantial and variable economic burden of rare diseases in South Carolina and suggest that targeted interventions may be necessary to address specific populations and conditions.

Table 3: The 11 Most Common RD Conditions Identified. Total Number of patients and Condition Prevalence per 1,000 SC Residents (year 2022)

ICD-10	Disease Name	Total	Prevalence
M350	Sjogren's syndrome	8868	1.650
G7000	Myasthenia gravis not otherwise specified	3723	0.693
D45	Polycythemia vera	2858	0.532
G6109	Guillain-Barre syndrome	1885	0.351
D680	von Willebrand's disease	1388	0.258
E849	Cystic fibrosis unspecified	1372	0.255
E8801	Alpha-1 antitrypsin deficiency	1184	0.220
Q612	Polycystic kidney disease	1104	0.205
M34.9	Systemic sclerosis unspecified	1047	0.195
G1221	Amyotrophic lateral sclerosis	566	0.105
G7101	Duchenne syndrome	238	0.044

Table 4: Total hospital admissions, ED visits and outpatient surgery/diagnostic procedure used from 2018 through June 2024 by the 196,799 patients included in the 38 RD conditions identified

	Pediatric Records for 13 RD Diagnoses 2018-2024	Adult Records for 26 RD Diagnoses 2018-2024	Totals Peds and Adult RDs 2018-2024	Per Year over 6.5 Years in Study
Total Cases	38,313	158,486	196,799	NA
Total Events	510,375	1,551,710	2,062,086	317,244/year
Hospital Admissions	89,358	374,948	464,096	71,399/year
Hospital Days	49,148	2,128,875	2,178,023	335,080/year
Estimated* Hospital Cost	$1.48 Billion	$7.37 Billion	$8.85 Billion	$1.36 Billion/year
ED Visits	338,057	667,957	1,006,014	154,771/year
Estimated* ED Cost	$526.1 Million	$1,39 Billion	$1.92 Billion	$294.8 Million/year

*Assumed 27% Cost-to-charge ratio

4. Expense Report

	FY2025 NCE 07/01/25 – 10/31/25	FY2026 11/01/25 – 6/30/25	Total
Salaries	$45,077	$106,271	$151,347
Fringe	$20,329	$48,031	$68,360
Symposium and Outreach	$0	$24,624	$24,624
Data Analytics	$15,000	$0	$15,000
Annual Report Postage	$49	$0	$49
Travel	$3,331	$540	$3,871
	$83,785	**$179,466**	**$263,251**

Appendix A. SC Rare Disease Symposium Agenda

South Carolina Rare Disease Symposium
Friday, February 27, 2026

9:30 AM **Check-In and Registration**

10:00 AM **Welcoming 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)

10:20 AM **South Carolina Rare Disease Prevalence Data**
Patrick Flume, MD

10:45 AM **Million-Dollar Treatments, Finite Budgets: Health Economics and Fair Choices in Rare Diseases**
Brian Chen, JD, PhD, Associate Professor, Health Services Policy and Management, Arnold School of Public Health, University of South Carolina (USC)

11:20 AM **Transition to Adulthood: Non-Medical Issues That Impact Lives of People with Rare Diseases**
David Rotholz, PhD, Executive Director and Clinical Professor, Center for Disability Resources (UCEDD, LEND), USC School of Medicine, Columbia

12:10 PM **Lunch**

12:40 PM **South Carolina Resources Panel**
Moderator: Neena Champaigne, MD, Medical Geneticist, Clinical Professor of Pediatrics, Division Chief, Medical Genetics and Genomics, MUSC

Panelists:
Dori Tempio, MS, Senior Director of Community Education, Able SC
Kathryn Padgett, Chief Operating Officer, Family Connection of SC

1:15 PM **Patient and Provider Perspectives**
Moderator: Neena Champaigne, MD

Panelists:
Sarah Mennito, MD, MSCR, Professor of Pediatrics and Internal Medicine, MUSC
Kate Wink

2:00 PM **Adjourn**

Appendix B. Estimations of Rare Disease Prevalence in SC

Kit N Simpson, DrPH
Professor
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ABSTRACT

This draft report presents an estimation of rare disease (RD) prevalence in South Carolina (SC) for 38 specific diagnoses selected from those with highest prevalence listed in the Artia Solutions RD Summary Inventory of RD conditions and their associated ICD-10 codes, provided by Genetech.

We used data from the SC All-payer data set from 2018 to mid-2024 to estimate prevalence based on hospital admissions and emergency department (ED) visits. We adjusted the prevalence estimates to capture patients with only medical office visits using Medicare and Commercial insurance data. Total number of hospital admissions, hospital days, ED visits, and the total cost to the health system were estimated from recorded charges using a 27% cost-to-charge ratio. Annual healthcare costs per patient, from a payer perspective for each RD condition, were estimated from SC Medicare payment records from older patients. Costs per patient for younger patients were based on commercial insurance records.

The top 38 prevalent RD conditions examined include a total of 29,300 unique SC residents. SC county RD rates varied from 0.04-1.65 RD patients/1000 population. Higher rates occur in counties with greater medical resources, suggesting RD under-diagnosis associated with low provider density. In 2023, these RD patients had 13,000 hospital admissions, using 76,500 hospital days, and 25,000 ED visits. Overall cost for this care was $490 million in 2024. Mean estimated annual cost per person for the RD conditions range from a low of about $8,000/year to a high of greater than $200,000/year. Thus, the economic burden of RD in SC varies greatly, indicating that interventions may need to be targeted to specific populations or conditions.

INTRODUCTION

The prevalence of rare diseases (RD) for the state of South Carolina (SC) is not known. This makes it difficult to estimate the services needed for these special populations. This exploratory study uses three data sources to develop and validate a method to identify RD condition in SC with a relatively high prevalence. The study objectives were to:

1. Develop a method of estimation of RDs for SC
2. Test the method in several available databases
3. Estimate SC population prevalence numbers for about 40 conditions
4. Estimate medical care use and economic burden to patents and payers

We report on the methods developed, the data used to apply our analyses including their limitations, and the findings below.

METHODS

This is an archival data analysis aimed at identifying patients with high prevalence RD in SC from medical care billing records for the state. Two data sources were used for each RD estimation. We use the Atria defined ICD-10 code(s) to extract inpatient hospital admissions (IP) and emergency department (ED) visits from data for 2018-2024. We then calculated the percentage of patients identified in IP, ED, and both IP and ED. Because the SC All-payer database does not contain records for medical office visits, we used a second data source to re-estimate IP and ER visits and calculate the percent of RD cases not included in the IP and ER data. We then estimated the number of patients for each RD for the state based on the two different data sources.

Data Sources

The major source of data used was the SC All Payer database (RFA data). The sources of these data are all bills for hospital admissions, emergency department visits and outpatient surgeries that are provided by acute care hospitals and outpatient surgery centers in the state. These archival billing data are de-identified to preserve patient and provider confidentiality. The variables captured are based on data points required for filing the 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 disease that includes a 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 secondary diagnosed conditions. The RFA data does not capture visits in ambulatory settings, such as a medical office, Federally Qualified Health System (FQHS) or nursing home. To estimate the number of patients who used only care in ambulatory settings (i.e. those that are missed in the RFA data source), we used two other databases which were limited to patients with Medicaid coverage or patients covered by commercial insurance. These databases are specific for insured patients, containing only a sample of the population and have age limitations. We used the MarketScan Commercial Insurance data set (MSC data) for patients up to age 65 years to capture the percentage of patients with the RD who had no hospital, ED or OP surgery visits in 2022. We replicated the analysis performed in the RFA data and then identified any additional RD patients who had only medical office visits in 2022. For the population of older patients with Medicare coverage, we used the

2022 Medicare 5% Limited Data set (MC) to estimate those who may be missed when using only RFA data.

Data Analysis

We first extracted all records for SC patients with the rare disease (RD) conditions of interest from the RFA data. The conditions of interest were selected from those with highest prevalence listed in the Artia Solutions RD Summary Inventory of RD conditions and their associated ICD-10 codes, provided by Genetech. We included patients with ICD-10 codes 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, ED and OP episodes recorded in the RFA data from 2022. We then extracted all records for patients with an RD diagnosis from the hospital admission and outpatient care files for the MSC data and the MC data sets 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 denominator population) what were included in the MSC and MC data sets. This is the underlying age and insurance type -specific population that was the source of the RD events. We used SC census data combined with the RFA population data on age and insurance to calculate the RD rates for SC. For the cost data estimation, we constructed a file of individual RD patient IDs and used this “finder” file to extract all their payment records for all healthcare services provided for one calendar year. We aggregated payments for services at the individual patient level and reported the mean payment (cost) of the care received.

RESULTS

Demographics, Prevalence and SC Rates

The results of the analyses are presented in the tables below. The prevalence and demographics of patients with RD in SC are shown in Table 1 and Figure 1. Table 2 lists the number and rates per 1,000 residents of the 11 most common RD conditions identified.

Table 1: Population Demographics for the 38 RD Conditions (year 2022)

	Variables	Number (%) N = 29,300	Statistic
Age:	Less than 20	1,575 (5.4)	p<.0001
	20 to 65	13,686 (46.7)	
	65 and Older	14,039 (47.9)	
Sex:	Male	10,716 (36.6)	p<.0001
	Female	18,584 (63.4)	
Race:	White	22,042 (75.2)	p<.0001
	Black	5,941 (20.3)	
	Other	1,317 (4.5)	
Insurance:	Medicare	15,929 (54.4)	p<.0001
	Private	7,401 (25.3)	
	Medicaid	2,675 (9.1)	
	Other	3,295 (11.3)	

Table 2: The 11 Most Common RD Conditions Identified. Total Number of patients and Condition Prevalence per 1,000 SC Residents (year 2022)

ICD-10	Disease Name	Total	Prevalence
M350	Sjogren's syndrome	8868	1.650
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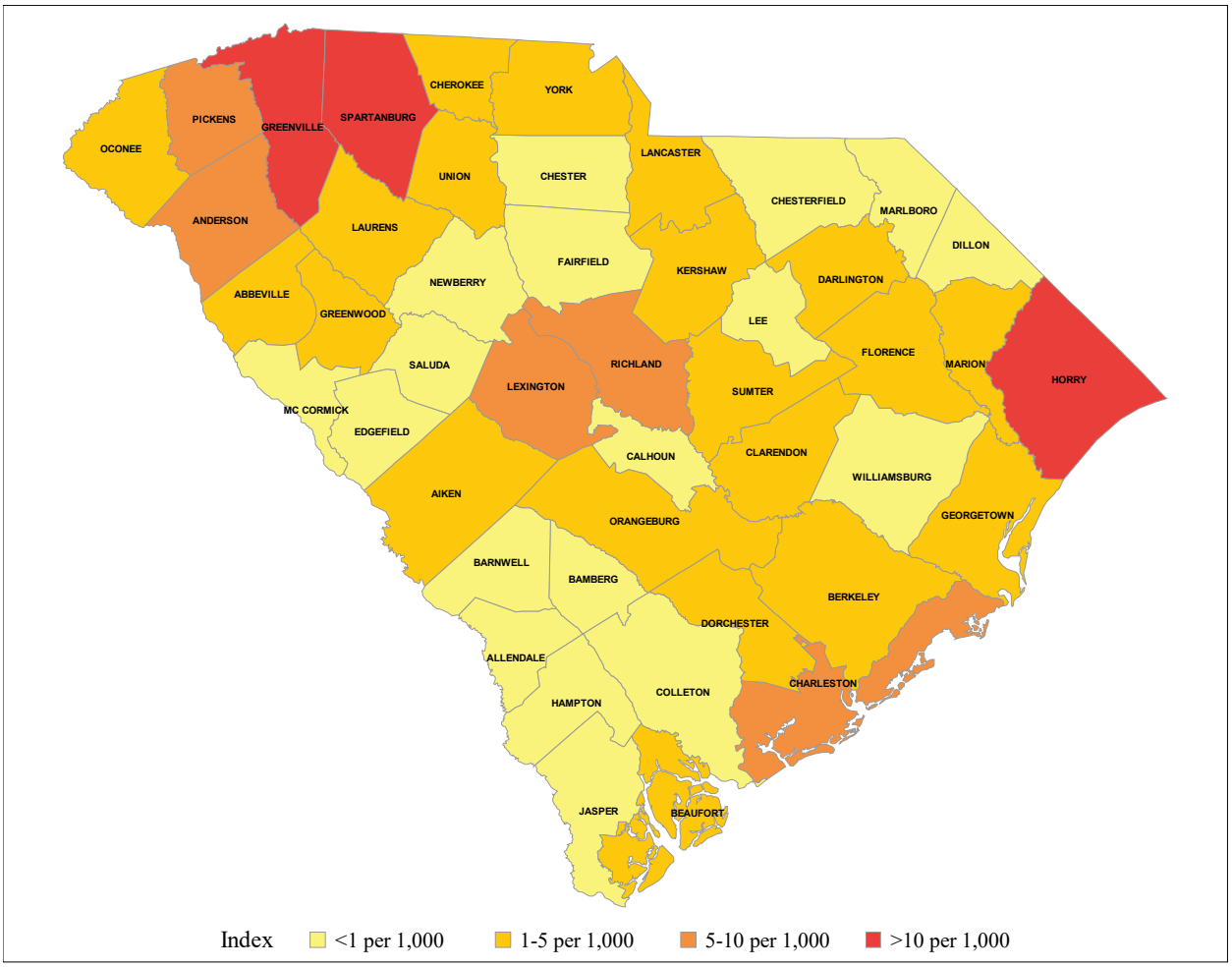
Q612	Polycystic kidney disease	1104	0.205
M34.9	Systemic sclerosis unspecified	1047	0.195
G1221	Amyotrophic lateral sclerosis	566	0.105
G7101	Duchenne syndrome	238	0.044

Figure 1: Distribution of RD Patients by County (rate per 1,000 county residents)



Results for the Hospital and ED Use 2018-2024 and Estimated Total Cohort Hospital-based Costs

The 38 RD conditions examined for SC identified a total of 196,799 individuals in the state with at least one RD event between 2018 and June 31, 2024. Of these individuals, 19.5% were under age 50 and 80.5% were 50 or older at the time of identification. Patients were found across the study years with between 9% and 16% identified for the first time in each calendar year. At the time of first diagnosis (in these data), 65% were Medicare beneficiaries, 8% were covered by Medicaid (mostly younger patients), 28% had private/commercial insurance, and the balance was either covered by other payers or were uninsured.

A large proportion of records for RD patients do not include the RD condition in the diagnosis codes recorded. Thus, summing the records with the RD diagnoses will greatly underestimate the medical care use by this population. To overcome this issue, we constructed a “Finder File” for the 196,899

patients and extracted all their records from the SC RFA data base used to calculate the resources resulted in a total of 2,062,086 records of IP, ED and outpatient surgery files for the populations defined by the 38 RD ICD-10 codes. A summary of the resources and the estimated cost of the care used by the pediatric RD and older group RDs are provided in Table 3 below.

Table 3: Total hospital admissions, ED visits and outpatient surgery/diagnostic procedure used from 2018 through June 2024 by the 196,799 patients included in the 38 RD conditions identified

	Pediatric Records for 13 RD Diagnoses 2018-2024	Adult Records for 26 RD Diagnoses 2018-2024	Totals Peds and Adult RDs 2018-2024	Per Year over 6.5 Years in Study
Total Cases	38,313	158,486	196,799	NA
Total Events	510,375	1,551,710	2,062,086	317,244/year
Hospital Admissions	89,358	374,948	464,096	71,399/year
Hospital Days	49,148	2,128,875	2,178,023	335,080/year
Estimated* Hospital Cost	$1.48 Billion	$7.37 Billion	$8.85 Billion	$1.36 Billion/year
ED Visits	338,057	667,957	1,006,014	154,771/year
Estimated* ED Cost	$526.1 Million	$1,39 Billion	$1.92 Billion	$294.8 Million/year

*Assumed 27% Cost-to-charge ratio

Detailed Results for Individual RD Conditions

Next, we report results for the main analysis of the 38 RD conditions. Table 4 shows the prevalence of 21 RD categories for conditions where the majority of patients are older than age 50. The per-person cost estimates in this table are based on 2022 annual Medicare payments for SC patients with the condition. The ICD-10 diagnosis code used for the estimation is provided together with the name of the condition.

Table 5 shows data for 17 RD diagnoses for younger patients. The mean annual per person cost estimated for these conditions was based on payment records from private insurance data. Each RD condition has been assigned a study RD number for ease of tracking to the analytical code.

Table 4: 21 Rare Disease Prevalence for Older Populations Costs Estimated Using 2022 SC Medicare LDS 5% Sample Data

Rare Disease Name	**Observed ED and IP Cases SC All Payer Data**	**Total Annual Payment per Person by**	**Percentage of Cases Older Than Age 50**	**Data Source and ICD-10 Diagnosis Code Used**	**Number**

	Adjusted by Medicare Estimates	**Medicare 2022 Data**			
Lupus specified (profundo, panniculitis)	91	$8,241	69%	Medicare L93.2	11
Chronic lymphocytic leukemia B-cell	4,849	$15,115	97%	Medicare C91.10	13
Diffuse large B-cell lymphoma	3,067	$18,558	88%	Medicare C83.3	14
Amyloidosis	299	$47,259	93%	Medicare E85.81	16
Polycystic Kidney Disease	933	$55,515	66%	Medicare Q61.2	17
Sjogren's Syndrome	5,796	$10,463	77%	Medicare M35.0	18
Scleroderma	2,299	$13,410	78%	Medicare M34.9	19
Polycythemia Vera	1,648	$18,615	87%	Medicare D45	20
α 1 Antitrypsin Deficiency	855	$27,348	72%	Medicare E88.01	21
Amyotrophic Lateral Sclerosis	836	$25,948	89%	Medicare G12.2	22
Guillain Barre Syndrome	1,883	$44,366	64%	Medicare G61.0	23
Idiopathic pulmonary fibrosis	2,604	$28,170	97%	Medicare J84.112	24
Giant cell arteritis	1,643	$16,821	96%	Medicare M31.6	26
Thrombocytopenia purpura*	4,391	$30,866	65%	Medicare D69.3	28
Autoimmune hepatitis	1,732	$11,066	70%	Medicare K75.4	29
Primary biliary cirrhosis	898	$8,098	85%	Medicare K74.3	30
Buerger's disease	308	$84,744	68%	Medicare I73.1	31
Hemorrhagic telangiectasia	260	$10,267	68%	I78.0	32 1
Multiple myeloma no remission*	6,030	$39,580	94%	C90.00	33 4
Sclerosing cholangitis' primary	384	$8,684	54%	K83.01	34 8
Acute transverse myelitis	647	$48,704	60%	G37.3	35 10

Table 5: Seventeen Rare Diseases Estimated for Younger Population with Costs Based on Private Insurance Data

Rare Disease Name	Observed Cases Using 2028-2024 SC Data	Mean Cost per Patient Based on Medicare 2022 Data (SD)	Percentage of Cases Younger than Age 50	ICD-10 Diagnosis Code	Number
Other congenital malformations with skeletal changes	250	$69,176	68%	Q87.5	36
Fragile X syndrome	206	$22,614	74%	Q99.2	37
Duchenne Muscular Dystrophy	150	$89,206	85%	G71.01	38
Von Willebrand's Disease	1,204	$69,176	60%	D68.0	39
Cystic Fibrosis (all cases)	940	$212,691	77%	E84	5
Eosinophilic esophagitis	7,796	$20,106	69%	K20.0	40
Fabry disease	54	$199,134	65%	E75.21	41
Sickle cell disease (not rare)	5,084	$45,297	81%	D57.1	42
Sympathetic uveitis	1 case	$189,729	Peds	H44.139	43
Lennox-Gastaut syndrome	356	$85,405	88%	G40.81	2
Cystinuria	26	$48,778	73%	E72.01	3
Rett syndrome	96	$32,855	63%	F84.2	7
Osteogenesis imperfecta	460	$37,386	62%	Q78.0	9
Systemic Scleroderma (unspecified)	1,811	$39,856	78%	M34.9	15
Familial amyloidosis	13	$17,947 $22,788	69%	Medicare Market Scan E85.0	25*
Retinitis pigmentosa	304	$37,011	77%	H35.52	27

*Dual source estimate to check accuracy

Limitations of Methods and Data

We have developed and tested an approach to identifying the prevalence of RD in SC using the state's available data. We validated our computer code that uses ICD-10 codes applied to two separate data set for 6 years to estimate SC RD prevalence. However, estimates are subject to under-diagnosis, under-coding, and errors in submission of billing data. The most uncertainty lies in the pediatric conditions. They may be especially affected and should be re-estimated using SC Medicaid data to improve validity. We examined 40 RDs however due to poor ICD-10 code specificity, or inconsistent capture across data sources, two were excluded from this report. We also, by request, included sickle cell disease, even though it technically is not an RD.

ACKNOWLEDGEMENTS

The information presented here used de-identified data from the following sources:

Medicare 5% Limited Data Set (LDS) is the Centers for Medicare & Medicaid Services (CMS)

Merative MarketScan Data: Certain data were supplied by Merative as part of one or more Merative MarketScan Research Databases. Any analysis, interpretation, or conclusion based on these data is solely that of the authors and not Merative.

SC All-payer Data: This information is from the records of the South Carolina Revenue and Fiscal Affairs Office, Data Integration and Analysis Division. Our authorization to release this information does not imply endorsement of this study or its findings by either the Revenue and Fiscal Affairs Office or the Data Oversight Council.

Appendix C. SC RDAC Quarterly Meeting Minutes

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.

Membership:

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	A
Department of Public Health	Ginie Daguise, MSPH, PhD (SCDPH)	A
Department of Health and Human Services	Kevin Wessinger, MD (SCDHHS)	P
Greenwood Genetic Center	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, MPA (Santee Cooper)	P
South Carolina Primary Healthcare Association	Vicki Young, PhD (SCPHCA)	A
Biopharma Industry	Jonathan Hawayek (Genentech)	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

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 3rd 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

Membership:

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	X
Department of Public Health	Ginie Daguise, MSPH, PhD (SCDPH)	X
Department of Health and Human Services	Kevin Wessinger, MD (SCDHHS)	X
Greenwood Genetic Center	Steve Skinner, MD (President & CEO)	
Prisma Health - University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	X
South Carolina Hospital Association	Kate Wink, MPA (Santee Cooper)	X
South Carolina Primary Healthcare Association	Vicki Young, PhD (SCPHCA)	
Biopharma Industry	Jonathan Hawayek (Genentech)	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

Rare Diseases Advisory Council Quarterly Meeting
January 16, 2026
9:00 AM-10:00 AM

Meeting agenda:

- Welcome

- Planning for the 3rd Annual Rare Disease Symposium
  - General theme surrounding the different aspects of transition, including healthcare, independence, life skills and aging
  - Discussed different points of the agenda
    - RD Prevalence - will cover top 10 or 20 prevalent conditions
    - Health Economics 101 - for the community/lay audience; won't be specific to only rare diseases
    - Technical Assistance and Training
    - Resources Panel and Q&A - Able SC and Family Connection SC will share what they offer and how it impacts the rare disease community
    - Patient, Parent/Caregiver, Provider Panel
      - KW has patient and caregiver connections that might be helpful
      - Needs look different for more dependent adults and that might be valuable to address in part of the panels
      - Disabilities can be an important topic surrounding rare disease and could be a valuable element of future events

- Bill Tracking
  - General Assembly is back in session as of this week. In session every Tuesday, Wednesday and Thursday and that is when new bills can be proposed. It's an election year for the House and constitutional officers but not the Senate. We are in the second year of a 2-year legislative session. Any bills that have been submitted that haven't become law will be lost after this year and would have to be submitted again.
  - KW to update spreadsheet of tracked bills, reflecting first readings and any new introductions relevant to rare diseases. No committee meetings scheduled for any of the bills at this time but KW to keep checking and let the RDAC know when something is up for discussion.
    - LCI committee stands for Labor, Commerce & Industry

        - Mark Sweatman can help the RDAC to get before the committees to share information
    - May be helpful to develop a website page to share bill information and general advocacy resources, including disclaimers and links to help users find their representatives.
        - Necessary to include disclaimer language that says that the RDAC is just providing information and not advocating for anything in particular.
        - Necessary to establish a plan for regularly updating the page to ensure accuracy as bills are introduced, amended, or removed; include language that mentions that this is not an exhaustive list.
        - TP and KB to identify other RDAC websites that effectively present resources for reference in developing the new page.

- Responding to requests received to our RDAC email
    - There has been an increase in the volume and complexity of inquiries received. The group discussed the need for a standardized response process and the formation of a subcommittee to address these challenges.
        - Creating stock responses
            - How are other RDACs triaging messaging?
            - Free text v. drop-down message selections (limit free text to prevent oversharing of PHI)
            - Could there be an office at universities that field questions like this?
            - Risk management (legal review of statements)
        - Triaging potential resources for inclusion on the website
            - Potential resources to recommend:
                - NORD Information & Resources Services Team
                - Able SC
                - Family Connections of SC
                - SC Rare
    - Tracking data related to incoming messages would be a good metric to report on over time.
    - JH, KK interested in the subcommittee
        - Outlined the need for broader participation in the subcommittee; suggested involving external stakeholders such as legal, risk management, and state agencies
        - TP and KB to provide summary of types of emails that our coming through the request box.

- Adjourn

Membership:

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	X
Department of Public Health	Ginie Daguise, MSPH, PhD (SCDPH)	
Department of Health and Human Services	Kevin Wessinger, MD (SCDHHS)	X
Greenwood Genetic Center	Steve Skinner, MD (President & CEO)	
Prisma Health - University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	

South Carolina Hospital Association	Kate Wink, MPA (Santee Cooper)	X
South Carolina Primary Healthcare Association	Vicki Young, PhD (SCPHCA)	X
Biopharma Industry	Jonathan Hawayek (Genentech)	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)	X
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	X
Program Coordinator	Tara Pittman Kimberly Brown	X X

Rare Diseases Advisory Council Quarterly Meeting
April 10, 2026
9:00 AM-10:00 AM

Meeting agenda:

- Welcome

- Rare Disease Symposium Debrief
  - The symposium had 130 attendees, evenly split between online and in-person participation, marking a slight increase from the previous year.
  - The event received positive feedback regarding its overall impact and participants' likelihood to attend future events.
  - Dr. Chen's presentation on healthcare economics was received positively, as well as Dr. Simpson and Dr. Flume's presentation on state incidence data.
    - Dr. Simpson continues to refine incidence data for the top 100 rare disease conditions, addressing data limitations, collaboration with experts, and plans to incorporate additional datasets
      - KK suggested Dr. Silver as a potential connection.
  - While the symposium intentionally covers broad topics to avoid singling out specific rare diseases, some attendees expressed a desire for more condition-specific content.
  - Feedback related to Segra Park focused on audio/visual and logistical concerns, including problematic backlighting and changes in parking arrangements. The group briefly discussed whether other venues in the Columbia area could be explored.
    - VD suggested Health Campus auditorium
  - KK observed that the symposium fostered cross-engagement among attendees and suggested that motivated participants could be invited to join subcommittees for future initiatives.
  - TP responded to a chat question by confirming that advocacy groups are invited to attend and may be offered table space at future events, provided a fair vetting

process is established. The team is open to increasing shared resources and engagement with advocacy organizations.
    - NC noted that the symposium sometimes coincides with national rare disease events and with other state-level meetings. The team discussed the need to consider timing to maximize attendance and representation.
        - Karen suggested highlighting concurrent rare disease activities across the state during the symposium to raise awareness and foster a sense of broader community engagement, even when scheduling conflicts are unavoidable.
    - The group reviewed the challenges of securing venues and organizing content for future symposiums, discussed funding timelines, and emphasized the need for earlier support from council members in identifying speakers and topics.

- Planning for the Annual Report
    - The annual RDAC report is due to the legislature in June. The group outlined plans for the upcoming annual report, including highlighting symposium outcomes, incidence data initiatives, participation in the NORD Breakthrough Summit, improvements in managing community outreach, and policy engagement.

- What's Next for SC RDAC?
    - NC discussed opportunities for collaboration with other southeastern RDACs, including the possibility of convening at the Southeastern Regional Genetics Group meeting to share policies and resources, and the group considered the benefits of cross-state engagement for rare disease communities.

- Old Business
    - Website Management
        - Responding to requests received to our RDAC email
        - Policy Webpage Updated
        - The group discussed the current structure and limitations of the council's website, addressed feedback on resource accessibility for caregivers and families, and invited suggestions for improving the site's organization and usability.

- Adjourn

Membership:

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	-
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Department of Health and Human Services	Kevin Wessinger, MD (SCDHHS)	P
Greenwood Genetic Center	Steve Skinner, MD (President & CEO)	P
Prisma Health - University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	P
South Carolina Hospital Association	Kate Wink, MPA (Santee Cooper)	-
South Carolina Primary Healthcare Association	Vicki Young, PhD (SCPHCA)	-
Biopharma Industry	Jonathan Hawayek (Genentech)	-

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	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)	P
Program Coordinator	Tara Pittman Kimberly Brown	P P