Rare Disease Advisory Council

Annual Report

Fiscal Year 2023-2024

July 1, 2024

Submitted to:

Ron DeSantis
Governor

Joseph A. Ladapo, MD, PhD
State Surgeon General

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Executive Summary

Annually, the Florida Department of Health (Department) is to submit a report to the Governor and the State Surgeon General which describes the activities of the Rare Disease Advisory Council (RDAC) in accordance with section 381.99, Florida Statutes. The report describes RDAC activities from the past year and its findings and recommendations regarding rare disease research and care in Florida.

The 2024 RDAC annual report, presented by the Department, underscores the collaborative efforts between the Department and the RDAC to address the needs of individuals with rare diseases.

Established by law in 2021 (Chapter No. 2021-122), the RDAC was created adjunct to the Department. The RDAC is composed of representatives from state agencies, health care providers, researchers, advocacy groups, insurance, and pharmaceutical industries, as well as individuals with rare diseases and caregivers of individuals with rare diseases. RDAC members hold a shared vision: To improve health outcomes for individuals residing in Florida who have rare diseases.

In 2023 the RDAC and the state of Florida have made several key advancements to understand the burden and costs associated with rare diseases in the state:

- RDAC members reviewed data from Florida Medicaid to identify the top 50 rare disease among those recipients.
- Reviewed the overlap of the top 50 rare diseases with the list of conditions included on the Florida Newborn Screening Program to support early detection and intervention for infants with those conditions.
- Made recommendations to create a comprehensive resource repository for patients, families and health care providers, which when implemented, will improve access to information and support.

These accomplishments demonstrate the RDAC's commitment to addressing and raising awareness of the unique needs of the rare disease community through targeted strategies and collaborative efforts.

June 13, 2024

The Honorable Ron DeSantis
Governor of the State of Florida
The Capitol – Plaza Level 05
400 South Monroe Street
Tallahassee, Florida 32399-0001

Dr. Joseph A. Ladapo, MD, PhD
State Surgeon General
4052 Bald Cypress Way
Tallahassee, FL 32399

Dear Governor DeSantis and Surgeon General Ladapo:

The Rare Disease Advisory Council was established in section 381.99, Florida Statutes, to assist the Department of Health by providing recommendations to improve health outcomes for individuals and their families residing in the state who are impacted by a rare disease.

On behalf of the members of the Rare Disease Advisory Council (Council), it is my pleasure to submit the 2024 annual report as outlined in Florida Statutes. This report documents the efforts of the Council over the past year and outlines the Council's dedication to improving health outcomes for Floridians who have a rare disease.

While each rare disease is different, the obstacles faced by individuals living with a rare disease and their families are common. The Council seeks to identify barriers and address the unique needs of the rare disease community by improving access to care, education, and other essential resources.

Additionally, the Council seeks to encourage greater research on rare diseases in Florida and provide resources for individuals and families who are impacted by these conditions.

Sincerely,

Melissa Jordan, MS, MPH
Chair, Rare Disease Advisory Council
Assistant Deputy Secretary for Health Florida Department of Health

Council Membership

Council Chair:

- Melissa Jordan, MS, MPH - Assistant Deputy Secretary for Health, Florida Department of Health (Governor Appointment)

Council Vice Chair:

- Kathryn Hebda, MM - Chancellor, Florida College System (Governor Appointment)

Members:

- Adam Anderson - Caregiver, State Representative (Senate Appointment)
- Eric Biernacki - Caregiver (House Speaker Appointment)
- Barry Byrne, MD, PhD - Physician, Director, Powell Gene Therapy Center (Senate Appointment)
- Ann Dalton, MM - Bureau Chief, Agency for Health Care Administration (Governor Appointment)
- Anita Davis, PT, DPT, FNCP, CNPT - Physical Therapist, Brooks Rehabilitation (Senate Appointment)
- Rebekah Dorr – Director, Clinical Patient Advocacy (House Speaker Appointment)
- Zana Dupee, JD - Caregiver (Senate Appointment)
- Jonathan Hawayek, MBA - Head of State Government Affairs, Spark Therapeutics, Inc. (Governor Appointment)
- Scott McClelland, PharmD - Vice President at Florida Blue (Governor Appointment)
- Jessica O'Reilly, JD - Self-Advocate (House Speaker Appointment)
- Divya Patel, DO, MBA - Program Director, University of Florida (Senate Appointment)
- India Steinbaugh, MPH - Self-Advocate (Senate Appointment)
- Jennifer Sutherland - Caregiver (House Speaker Appointment)
- Mustafa Tekin, MD - Division Director, University of Miami (House Speaker Appointment)
- Rajan Wadhawan, MD, MMM - Senior Executive Officer, AdventHealth (House Speaker Appointment)

Vacant Positions:

- Representative of Office of Insurance Regulation
- Pharmacist with rare disease experience
- Geneticist
- Registered nurse with rare disease experience
- Hospital administrator from a hospital providing care to rare disease patients

Background

Rare diseases pose unique challenges due to their limited prevalence and often complex nature. These conditions often result in significant physical, emotional, and financial burdens for individuals and their families.

The Orphan Drug Act defines a rare disease as a disease or condition that affects fewer than 200,000 people in the United States.[1] Estimates suggest roughly 10% of the U.S. population are living with a rare disease or condition.[2] This estimate has been used by the rare disease community for several decades to highlight that while individual diseases may be rare, the total number of individuals living with rare diseases is large. In a recent study estimated 15.5 million children and adults were affected by 379 rare diseases in 2019. The total economic burden associated with these rare diseases amounted to a staggering $997 billion[3].

In Florida, it is estimated that as many as 2.2 million people may be impacted by rare diseases, including conditions such as phenylketonuria, Rett's syndrome, and Hereditary factor IX deficiency, and more recognizable rare conditions like cystic fibrosis and sickle cell disease.

Rare diseases encompass a diverse range of conditions, including genetic disorders, infectious diseases, cancers, and various other pediatric and adult conditions.[4] A rare disease can affect anyone at any point within the lifespan. Rare diseases can be acute or chronic and are characterized by the variation of symptoms not only displayed from one rare disease to another, but also among individuals who are diagnosed with the same disease. Although the features of specific rare diseases can differ in a myriad of ways, the effects on life and functioning are often similar for affected individuals and their families. Individuals who live with rare diseases may face reduced quality of life due to the lack or loss of autonomy caused by chronic, progressive, degenerative, and life-threatening aspects of many rare diseases.

Rare diseases have many different causes. It is believed that 80 percent or more of rare diseases are genetic.[5] For genetic rare diseases, genetic testing is often the only way a definite diagnosis can be made. Research shows genetic rare diseases are directly caused by a variant, or change, in a gene or chromosome. Variants can be inherited from a parent or occur during an individual's lifetime.

There are several rare diseases that do not have a known genetic cause. These include some types of infections, autoimmune diseases, certain forms of cancer, diseases caused by exposure to toxins or other environmental factors, and some conditions which have multiple possible causes.[6] For specific rare diseases that have been named and characterized for decades, investigators still have not determined a cause. However, many rare diseases with reasonably well understood causes and mechanisms still lack effective treatments or preventive strategies. Rare diseases present fundamentally different challenges from those of more common diseases.

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[1] Orphan Drug Act, H.R.5238, 97th Congress (1981-1982). (1983, January 4). Retrieved from https://www.congress.gov/bill/97th-congress/house-bill/5238

[2] U.S. Department of Health and Human Services. (2021, January 26). FAQs About Rare Diseases. Genetic and Rare Diseases Information Center. Retrieved from https://rarediseases.info.nih.gov/about

[3] Yang, G., Cintina, I., Pariser, A. et al. The national economic burden of rare disease in the United States in 2019. Orphanet J Rare Dis 17, 163 (2022). https://doi.org/10.1186/s13023-022-02299-5

[4] Rare Genetic Diseases. Genome.gov. (2018, April 13). Retrieved from https://www.genome.gov/dna-day/15-ways/rare-genetic-diseases

[5] Rare Genetic Diseases. Genome.gov. (2018, April 13). Retrieved from https://www.genome.gov/dna-day/15-ways/rare-genetic-diseases

[6] Field, M. J., & Boat, T. F. (2010). Profile of Rare Diseases. In Rare Diseases and Orphan Products: Accelerating Research and Development. essay, National Academies Press. Retrieved from https://www.ncbi.nlm.nih.gov/books/NBK56184/?msclkid=6d5bce28d07c11ec85146bea249c2fa3.

Individuals with undiagnosed genetic diseases often face an uncertain and unpredictable journey, referred to as a "diagnostic odyssey."

For one-third of individuals with rare diseases, getting an accurate diagnosis can take one to five years[7]. Individuals with rare diseases often seek treatment in health care settings where their condition has never been seen before. Many health care providers are often unfamiliar with the multitude of symptoms that can be associated with rare diseases. Additionally, absent, masked, misunderstood, or confusing symptoms can contribute to a delayed diagnosis.

[7] FDA.(2018, March 1). Orphan Products: Hope for People with Rare Diseases. U.S. Food and Drug Administration. Retrieved from https://www.fda.gov/drugs/information-consumersand-patients-drugs/orphan-products-hope-people-rare-diseases

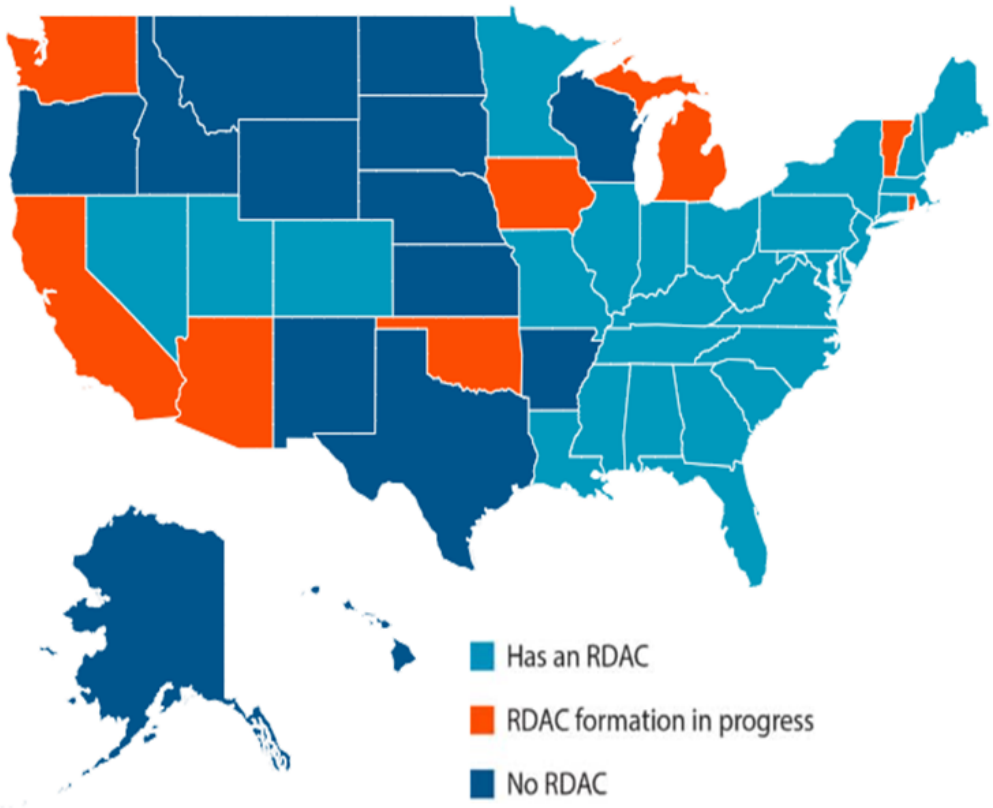
Introduction

In 2021, section 381.99, Florida Statutes was created, and established a Rare Disease Advisory Council (RDAC). Florida became the 19th of 23 states to create a rare disease Council.[8] (See Figure 1).

Florida's RDAC was created to provide recommendations to the State Surgeon General and the Governor on ways to improve health outcomes for individuals affected by rare diseases and their families. With a focus on improving treatment access and developing strategies to educate providers on how to recognize and diagnose rare diseases, the RDAC's work is a vital part of Florida's efforts to improve patient care across the health care system.

Further, achieving a timely and accurate diagnosis ensures that individuals living with rare diseases can receive the clinical care and counseling appropriate for their disease. A diagnosis also removes a significant psychological weight from those who have been struggling with a long "diagnostic odyssey." It allows individuals and their families affected by rare diseases the opportunity to connect with a rare disease community. This report provides a summary of the work performed, recommendations and solutions developed, to increase awareness, diagnostic tools, and access to affordable treatment and cures for those affected by rare diseases.

Figure 1: Rare Disease Advisory Councils by State.[9]



[8] NORD (National Organization for Rare Disorders). (2023, March 27). RDACs by State. Retrieved from https://rarediseases.org/policy-issues/rare-disease-advisory-councils/
[9] NORD. RDACs by State (2024). Retrieved from https://rarediseases.org/rare-disease-advisory-councils/map/

Functions of the Council

As outlined in section 381.99 (4)(a) through (d), Florida Statutes, the advisory council shall:

Consult with experts on rare diseases and soliciting public comment to assist in developing recommendations on improving the treatment of rare diseases in Florida.

Develop recommended strategies for academic research institutions in Florida to facilitate continued research on rare diseases.

Develop recommended strategies for health care providers to be informed on how to recognize and diagnose rare diseases to effectively treat patients more effectively. The advisory council shall provide such strategies to the Department of Health for publication on the Department's website.

Provide input and feedback in writing to the Department, the Medicaid program, and other state agencies on matters that affect individuals who have been diagnosed with rare diseases, including, but not limited to, pandemic or natural disaster preparedness and response.

Council Activities

Fiscal Year 2023-2024 Timeline

August 2023

- August 15: The Academic Research Institutions Subcommittee and the Health Care Providers Subcommittee convened for a joint meeting.
- August 17: The State Agencies Subcommittee convened.

September 2023

- September 19: The Academic Research Institutions Subcommittee convened.
- September 20: The Health Care Providers Subcommittee convened.
- September 21: The State Agencies Subcommittee convened.

October 2023

- October 25: The Council convened.

November 2023

- November 14: The Academic Research Institutions Subcommittee convened.
- November 15: The Health Care Providers Subcommittee convened.
- November 16: The State Agencies Subcommittee convened.

December 2023

January 2024

- January 23: The Academic Research Institutions Subcommittee convened.
- January 24: The Health Care Providers Subcommittee convened.
- January 26: The State Agencies Subcommittee convened.

February 2024

- February 7: The Council convened.

March 2024

- March 26: The State Agencies Subcommittee convened.
- March 26: The Academic Research Institutions Subcommittee convened.
- March 27: The Health Care Providers Subcommittee convened.

April 2024

- April 24: The Council convened.

May 2024

- May 16: The Academic Research Institutions Subcommittee convened.
- May 14: The Health Care Providers Subcommittee convened.
- May 15: The State Agencies Subcommittee convened.

FY 2023-2024 Accomplishments

Council Meeting Operations and Procedures

Since July 2022, the full RDAC has convened six times and continues to meet on a quarterly basis. The RDAC subcommittees meet monthly, except during months when the full RDAC meets.

RDAC Subcommittees

The RDAC established three specialized subcommittees to effectively fulfill the requirements outlined in section 381.99 (4) (b-d), Florida Statutes, (Appendix A), showcasing a robust and comprehensive approach to addressing the challenges of rare diseases. The three subcommittees are designated as follows:

1. **The Academic Research Institutions Subcommittee** champions collaboration and seeks to propel research initiatives within academic and research settings.
2. **The Health Care Providers Subcommittee** raises rare disease awareness and seeks to enhance diagnosis, and refinement of treatment strategies among health care professionals.
3. **The State Agencies Subcommittee** works to align efforts and policies across government agencies to provide better support for individuals with rare diseases.

Subcommittees meet with the goal of nurturing ongoing dialogue, collaboration, and tangible progress in meeting the unique needs of individuals affected by rare diseases. These meetings serve as platforms for stakeholders to share insights, discuss best practices, and devise targeted strategies aimed at improving health outcomes and enhancing the quality of life for this population.

Collaboration with Experts on Rare Diseases

RDAC members also collaborate with national and state experts on rare diseases to meet council goals. As a result, the RDAC has developed recommendations to improve rare disease treatment, enhance health care provider awareness, and facilitate timely diagnosis strategies.

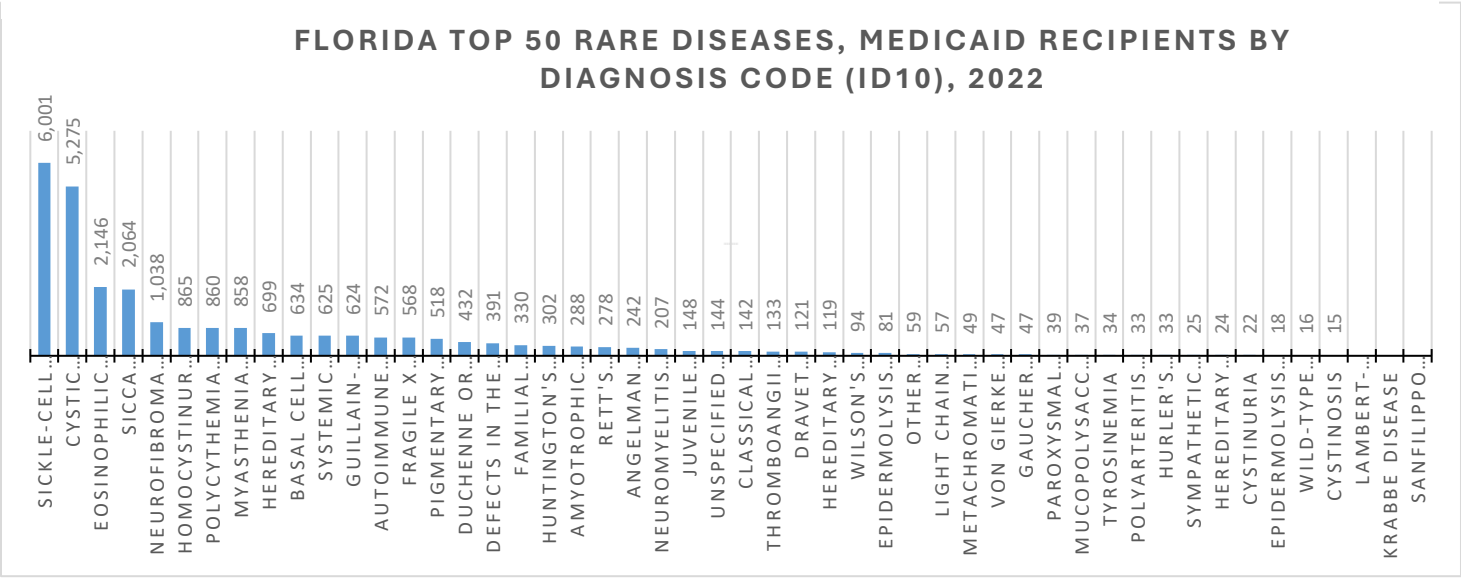
Identified 50 Most Common Rare Diseases in Florida Among Medicaid Recipients

By capturing hospital billing data, the RDAC has been able to identify the top 50 rare diseases for Florida Medicaid recipients. International Classification of Diseases (ICD)10 codes are alphanumeric codes used by health care providers to classify and code diagnoses, symptoms, and procedures for billing purposes and medical record documentation.

By analyzing hospital billing information containing ICD-10 codes, researchers and health care professionals can identify patterns and trends in disease prevalence and incidence. It is important to note that the Medicaid population may not be reflective of the state's population, but this data represents the best source currently available to the RDAC and the Department. Appendix B presents the top rare diseases, from 1018-2022 by diagnosis, captured using Florida Medicaid billing data.

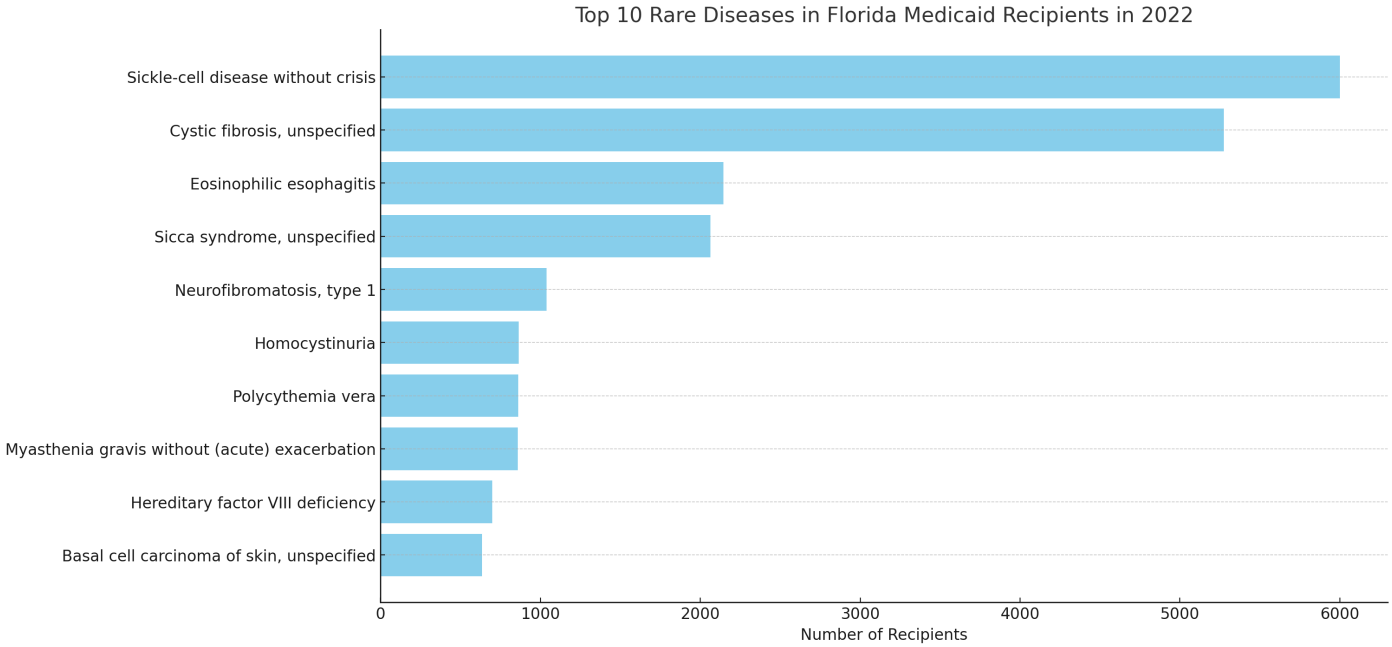
Figures 2 presents 2022 data on the Florida Top 50 Rare Diseases, by ICD-10 Code, and Figure 3 shows the Top 10 Rare Diseases as of 2022.

Figure 2: Florida Top 50 Rare Diseases, Medicaid recipients by ICD-10 Code*



Source: Florida's Agency for Health Care Administration.

Figure 3: Florida Top 10 Rare Disease, 2022



Source: Florida's Agency for Health Care Administration.

* Please note that Medicaid data may not fully reflect the demographic and economic differences within Florida's population. While it provides valuable insights, it primarily pertains to individuals enrolled in the Medicaid program and may not be representative of broader demographic and economic trends within the state.

Identified Rare Diseases Covered by Florida Newborn Screening Program

The Department received a list of the top rare diseases from the Genetic and Rare Diseases Information Center (GARD) as well as a list of conditions covered by the Florida Newborn Screening Program. The lists were compared by ICD-10 code to identify the rare diseases covered by the Florida Newborn Screening Program.

The Florida Newborn Screening Program screens for 58 conditions, 35 of which are core conditions and 23 are secondary conditions. Fifty-three of the conditions are included in the Recommended Uniform Screening Panel that is recommended by the U.S. Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children.[10]

The seven core conditions screened by the Florida Newborn Screening Program are considered rare diseases by GARD: (1) propionic acidemia (Prop), (2) tetralogy of Fallot (critical congenital heart disease), (3) carnitine uptake defect (CUD), (4) medium-chain acyl-CoA dehydrogenase deficiency (MCAD), (5) very long-chain Acyl-CoA dehydrogenase deficiency (VLCAD), (6) primary congenital hypothyroidism, and (7) sickle cell disease.

The following secondary conditions screened by the Florida Newborn Screening Program are considered rare diseases by GARD: (1) short-chain Acyl-CoA dehydrogenase (SCAD), (2) hemoglobin C and E conditions, and (3) various hemoglobinopathies.

Catalog Resources for a Repository of Information on Rare Diseases

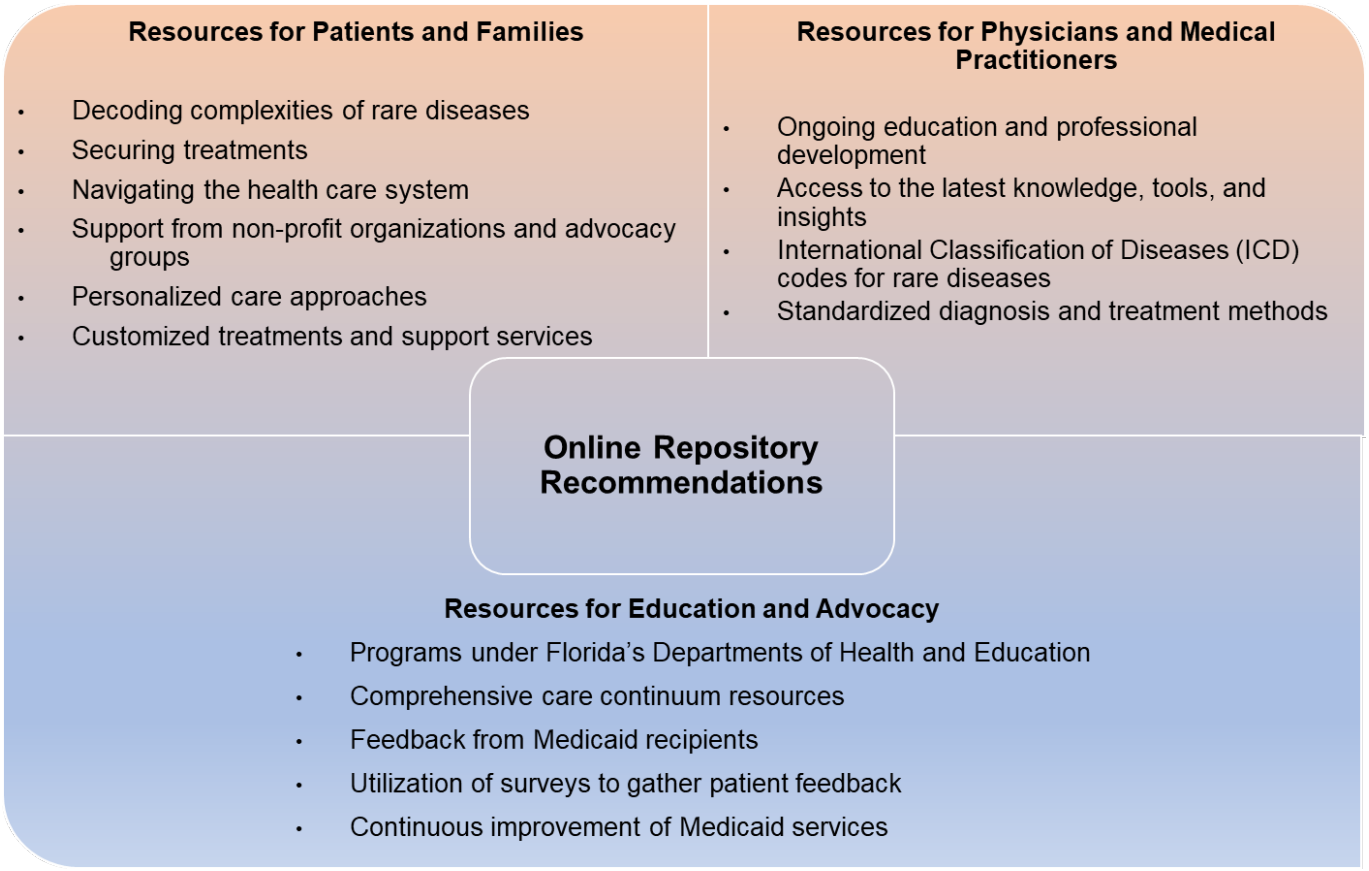
The RDAC has worked to catalog resources to support individuals, families, and health care practitioners dealing with rare diseases. This comprehensive repository (see Figure 4) includes tailored resources for families, ongoing education for health care providers, and connections to non-profit and advocacy groups.

The RDAC also emphasizes personalized patient care, offers standardized diagnostic and treatment approaches, and gathers feedback from Medicaid recipients to ensure continuous service improvement.

These efforts aim to establish a robust online repository to support health care providers and individuals impacted by rare diseases in Florida.

[10] Florida Newborn Screening: Conditions: What is Screened (2024). Retrieved from https://floridanewbornscreening.com/conditions/core-secondary-conditions/

Figure 4: Online Resource Repository Recommendations



FY 2023-2024 RDAC Recommendations

The RDAC plays an important role in advancing information on the treatment and care of individuals affected by rare diseases. By engaging experts and gathering public input, the RDAC makes recommendations geared toward enhancing rare disease treatment and promoting academic research while ensuring health care providers are equipped with the requisite knowledge to effectively identify and diagnose these conditions.

To enhance efficiency, the RDAC has three subcommittees: the Academic Research Subcommittee, the Health Care Providers Subcommittee, and the State Agencies Subcommittee. These subcommittees collaborate to generate ideas and provide input for the full RDAC's consideration.

The recommendations put forth by the subcommittees are reviewed and endorsed by the Council, ensuring a comprehensive approach to addressing the needs of rare disease-affected individuals and families in Florida (see Appendix C for full list).

Using the recommendations generated by the subcommittees, the RDAC identifies their priorities for the next fiscal year, Figure 5 shows the top five priorities endorsed by the RDAC.

Figure 5: Top Five RDAC Recommendations

1

Enhancement of Education and Awareness

- Promote education on rare diseases among health care providers, patients, families, and caregivers through centralized educational resources and expert consultations available on public websites.
- Develop and distribute resource guides and training materials for health care providers, school staff, and emergency personnel to improve understanding and support for individuals with rare diseases.

2

Enhanced Collaboration and Funding

- Continue partnering with NORD to raise awareness, secure funding, and drive policy changes.
- Establish a special designation for Florida Rare Disease Institutions.
- Broaden collaboration and encourage continued funding to increase access to genetic testing and rare disease research.

3

Service Improvement and Access

- Develop needs assessments and evaluate service gaps for the Department of Health, Agency for Health Care Administration, and Office of Insurance Regulation to enhance services for individuals with rare diseases.
- Promote the development of state and Medicaid programs, improve insurance benefits, and ensure non-discriminatory practices toward individuals with rare diseases.

4

Data Collection and Research Enhancement

- Identify and define data needs to better understand Florida's rare disease population, including prevalence, incidence, and distribution across the state.
- Support research institutions by identifying best practices, promoting expanded access to clinical trials and genome sequencing, and encouraging continuous funding for research programs.

5

Emergency Preparedness and Response

- Conduct comprehensive needs assessments for disaster preparedness specific to the rare disease population.
- Develop and disseminate educational materials on emergency preparedness and ensure access to community resources during emergencies.

These recommendations reflect the RDAC's commitment to fostering a supportive and responsive environment for individuals with rare diseases in Florida, enhancing their quality of life, and ensuring equitable access to necessary services.

Conclusion

Florida's RDAC is committed to enhancing treatment and care for individuals with rare diseases. This report, developed through extensive collaboration and stakeholder engagement, offers key recommendations designed to improve diagnosis, treatment, and research of rare diseases. It emphasizes the need for standardized diagnostic procedures and a comprehensive analysis of the resources available to those affected.

Since its establishment, the RDAC has actively worked to refine its recommendations, urging stakeholders to transform these ideas into effective actions. The insights gained over the past year provides a strong foundation for future advancements, underscoring the RDACs dedication to supporting affected individuals and families in Florida. The successful implementation of these improvements relies on the collective effort of all stakeholders.

Appendices

Appendix A – Subcommittee Membership

Subcommittee/Members		
Academic Research Institutions	**Title/Position**	**Affiliation**
Barry Byrne, MD, PHD	Director, Powell Gene Therapy Center Associate Chair, Department of Pediatrics Professor, Pediatrics and Molecular Genetics & Microbiology	University of Florida
Divya Patel, DO, MBA	Program Director, Interstitial Lung Disease Program Director, Sarcoidosis Associate Professor of Medicine	University of Florida
Mustafa Tekin, MD	Division Director, Clinical and Translational Genetics	University of Miami
Adam Anderson	State Representative Caregiver	Florida House of Representatives
Rajan Wadhawan, MD, MMM	Senior Executive Officer AdventHealth for Children AdventHealth for Women	AdventHealth
Health Care Provider		
Anita Davis, PT, DPT, FNCP, CNPT	Physical Therapist	Brooks Rehabilitation
Rebekah Dorr	Director, Clinical Patient Advocacy	Myasthenia Gravis Hope Foundation
Zana Dupee, JD	Caregiver	
Jessica O'Reilly, JD	Self-Advocate	
India Steinbaugh, MPH	Self-Advocate	
Eric Biernacki	Caregiver	
State Agencies		
Ann Dalton	Chief, Bureau of Medicaid Policy	Florida Agency for Health Care Administration
Kathryn Hebda, MM	Chancellor, Florida College System	Florida Department of Education
Jonathan Hawayek, MBA	Director, State Government Affairs	Spark Therapeutics, Inc
Melissa Jordan, MS, MPH	Assistant Deputy Secretary for Health	Florida Department of Health
Scott McClelland, PHARMD	Vice President, Commercial and Specialty Pharmacy Programs and Health Care Solutions	Florida Blue
Jennifer Sutherland	Caregiver	

Appendix B – Rare Disease by Diagnosis in Florida 2018–2022

Florida Medicaid						
Total Number of Medicaid Recipients by Selected Diagnosis Codes						
(Service Date: 1/1/2018–12/31/2022)						
Diagnosis	ICD-10	Calendar Year				
		2018	2019	2020	2021	2022
Basal cell carcinoma of the skin, unspecified	C4491	1,045	772	716	742	634
Polycythemia vera	D45	756	742	760	838	860
Sickle-cell disease without crisis	D571	6,015	5,916	5,572	6,055	6,001
Paroxysmal nocturnal hemoglobinuria [Marchiafava-Micheli]	D595	48	31	34	44	39
Other constitutional aplastic anemia	D6109	84	60	58	59	59
Hereditary factor VIII deficiency	D66	743	580	596	662	699
Hereditary factor IX deficiency	D67	107	99	108	109	119
Severe combined immunodeficiency with reticular dysgenesis	D810					
Defects in the complement system	D841	328	304	301	313	391
Classical phenylketonuria	E700	392	327	236	206	142
Tyrosinemia	E7021	30	36	22	32	34
Cystinuria	E7201	22	18	16	20	22
Cystinosis	E7204	15	15			15
Homocystinuria	E7211	1,338	1,348	1,230	1,111	865
von Gierke disease	E7401	59	55	33	51	47
Gaucher disease	E7522	44	39	47	56	47
Krabbe disease	E7523	17	16	16		
Niemann-Pick disease type C	E75242					
Metachromatic leukodystrophy	E7525	47	50	54	50	49
Hurler's syndrome	E7601	33	34	25	33	33
Mucopolysaccharidosis, type II	E761	38	32	27	38	37
Sanfilippo mucopolysaccharidoses	E7622			19	15	
Hereditary erythropoietic porphyria	E800	34	49	36	20	24
Wilson's disease	E8301	95	85	76	99	94
Familial hypophosphatemia	E8331	298	271	260	317	330
Cystic fibrosis, unspecified	E849	1,195	1,143	1,071	1,472	5,275
Light chain (AL) amyloidosis	E8581	46	42	48	70	57
Wild-type transthyretin-related (ATTR) amyloidosis	E8582			15	15	16
Rett's syndrome	F842	301	270	267	262	278
Huntington's disease	G10	379	360	344	340	302
Amyotrophic lateral sclerosis	G1221	385	359	328	335	288
Neuromyelitis optica [Devic]	G360	153	164	169	214	207
Dravet syndrome, intractable, without status epilepticus	G40834			44	102	121
Guillain-Barre syndrome	G610	607	602	640	670	624
Myasthenia gravis without (acute) exacerbation	G7000	910	829	826	874	858

Lambert-Eaton syndrome, unspecified	G7080	15	18	18	15	
Duchenne or Becker muscular dystrophy	G7101	191	387	376	421	432
Unspecified hereditary retinal dystrophy	H3550	156	152	147	170	144
Pigmentary retinal dystrophy	H3552	600	486	412	488	518
Sympathetic uveitis, unspecified eye	H44139				17	25
Thromboangiitis obliterans [Buerger's disease]	I731	236	197	160	162	133
Eosinophilic esophagitis	K200	1,687	1,684	1,632	1,939	2,146
Autoimmune hepatitis	K754	643	584	564	569	572
Juvenile rheumatoid arthritis with systemic onset, unspecified	M0820	136	122	145	152	148
Polyarteritis with lung involvement [Churg-Strauss]	M301	39	30	34	44	33
Systemic sclerosis, unspecified	M349	662	598	603	634	625
Sicca syndrome, unspecified	M3500	1,811	1,689	1,709	2,059	2,064
Epidermolysis bullosa, dystrophic	Q812	23	26	28	22	18
Epidermolysis bullosa, unspecified	Q819	127	91	94	87	81
Neurofibromatosis, type 1	Q8501	968	942	915	988	1,038
Angelman syndrome	Q9351	93	242	230	242	242
Fragile X chromosome	Q992	587	653	524	556	568

Source: Agency for Health Care Administration Medicaid Data Analytics (RQ 4822) 2/3/2023
MDA SQL v claim and encounter tables as of 2/1/2023

If any of these results were less than 15, the total was suppressed for the privacy of the recipients with the diagnosis.

Appendix C – Recommendations and Strategies by Council Subcommittee

Academic Research Institutions Subcommittee

Academic Research Institutions Subcommittee	
Recommendation	**Strategy**
Support research institutions in Florida.	• Identify the components that constitute a best practice "Rare Disease Research Institution." • Elicit feedback on developed components for a best practice "Rare Disease Research Institution." • Centralize developed components of a best practice "rare disease research institution" on a publicly available website. • Identify and promote best practices established by rare disease research institutions on a publicly available website. • Encourage continued funding for the Pediatric Rare Disease Grant program.
Promote the availability of research institutions to individuals with rare diseases.	• Promote expanded access to genome sequencing for all individuals with rare diseases. • Establish a maximum waiting period to promote timely access to research clinical trials for individuals with rare diseases. • Promote timely access to research clinical trials for individuals with rare diseases. • Develop a database of research institutions that are available within a geographic location (ZIP code, city, region) on specific rare diseases. • Centralize resources on a publicly available website. • Create a community of practice amongst research institutions and individuals with rare diseases who are looking to be involved in research studies.

Health Care Providers Subcommittee

Health Care Providers Subcommittee	
Recommendation	**Strategy**
Promote education on rare diseases to health care providers.	• Identify existing educational resources, including curriculum in medical and nursing schools, and continuing education courses for health care providers on various topics related to rare diseases, including resources on patient-centered care and holistic medicine. • Centralize educational resources on a publicly available website.

Promote advancements in the process of achieving a diagnosis for rare diseases.	• Identify screening tools for health care providers on rare diseases. • Develop resources, such as infographics for health care providers upon suspicion of a rare disease through the process of referral, diagnosis, and treatment. • Develop resources, such as infographics for individuals with rare diseases and their families that explains referral, diagnosis, and treatment process for rare diseases.
(Combined with Academic Institutions) Promote Education on Rare Diseases and Enhance Support Resources.	• Enhance education about rare diseases for patients, families, caregivers, and health care providers. • Identify and publicize current medical practices and specialists who treat both pediatric and adult individuals with rare diseases. • Survey individuals with rare diseases to understand barriers to accessing care, including testing and treatment, and identify challenges faced by medical practices in accepting such patients. • Develop comprehensive resources for individuals with rare diseases on self-advocacy during health care interactions and for mental and emotional support. Ensure these resources also include guidance for long-term aspects like pain management, household needs, workplace and lifestyle adjustments, and overall quality of life. • Promote the inclusion of advocates, case managers, and interdisciplinary care teams to assist in facilitation during health care interactions. • Identify state and national rare disease experts for consultations with health care providers on complex cases and explore the use of technology to facilitate these consultations. • Centralize all consultation resources and patient advocacy groups that support the rare disease community on a publicly available website, making it easy for both patients and providers to access needed information.

State Agencies Subcommittee

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Recommendation	**Strategy**
Identify data needed to understand the population with rare diseases in Florida.	• Define the prevalence/ incidence of rare diseases in Florida. • Develop a list of rare diseases and identify the distribution of incidents across the state, beginning with the top 50 most prevalent rare diseases. • Identify Florida specific data from national rare diseases using the current available databases in Florida. • Determine where Florida falls in comparison to other states regarding rates of rare diseases.

Identify areas of improvement for the Florida Department of Health on rare diseases.	• Identify the population with rare diseases the Department serves. • Develop a needs assessment survey on the population with rare diseases the Department serves. • Identify barriers and gaps in services that impacts individuals with rare diseases that are serviced by the Department. • Publish data on rare diseases through the Department's data portals, such as FLHealthCHARTS.org • Develop training for Department staff to better serve the population with rare diseases. • Develop an evaluation plan.
Identify areas of improvement for the Florida Agency for Health Care Administration (AHCA) on rare diseases.	• Identify Medicaid data that can be used to determine the incidence/prevalence of rare diseases in Florida. • Identify Medicaid programs that serve the population with rare diseases. • Identify the population with rare diseases in Florida Medicaid serves and assess its needs. • Identify how current surveys are being utilized to collect feedback from Medicaid recipients with rare diseases in relation to receipt of Medicaid services. • Research potential opportunities to improve Medicaid programs or services and address barriers and gaps. • Develop training for AHCA staff to better serve Medicaid recipients with rare diseases. • Develop an evaluation plan.
Identify areas of improvement for the Florida Department of Education (DOE) on rare diseases.	• Develop a resource guide to educate public and private school staff, students with rare diseases, and families on available resources and supports such as Individualized Educational Plans, 504 Plans, and scholarships offered. • Develop a communication plan to include the distribution of available resources and supports to meet the needs of students with rare diseases and their families. • Develop training for DOE staff to better serve the population with rare diseases.
Identify areas of improvement for the Florida Office of Insurance Regulation (OIR) on rare diseases.	• Identify what data on rare diseases currently exist through commercial health insurance plans. • Identify the population with rare diseases OIR serves and assess its needs. • Identify if current surveys are being utilized to collect feedback from health insurance recipients with rare diseases. • Identify OIR programs that serve the population with rare diseases. • Evaluate requirements for insurance benefits not to be discriminatory against insurance recipients with rare diseases. • Research potential opportunities to improve health insurance programs or services and address barriers and gaps. • Develop training for OIR staff to better serve health insurance recipients with rare diseases.

Identify areas of improvement for disaster preparedness and pandemic response for individuals with rare diseases in Florida.	• Complete a needs assessment of the populations with rare diseases in Florida for natural disaster preparedness and pandemic response. • Identify local community resources individuals with rare diseases may contact in the event of an emergency for assistance. • Develop educational and training materials on the needs of individuals with rare diseases for emergency shelter coordinators and staff. • Develop educational materials for individuals with rare diseases on emergency preparedness planning to be shared with partner agencies, such as information on the Florida Special Needs Shelter Registry, and importance of having a checklist for individuals with rare diseases that highlights specific items they may need during a disaster.