RARE DISEASE ADVISORY COUNCIL

Fiscal Year 2024-25 Annual Report

July 1, 2025

Ron DeSantis
Governor

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State Surgeon General

Contents

Executive Summary

In the United States, rare diseases are defined under the Orphan Drug Act as medical conditions affecting fewer than 200,000 individuals annually. Despite their individual rarity, the collective impact of these disorders is significant, with estimates suggesting that approximately 2.2 million residents in Florida alone may be affected by one or more rare diseases. This underscores the critical need for continued research, policy development, and community support to address the challenges faced by individuals with rare diseases and their families.[1]

Pursuant to section 381.99, Florida Statutes (F.S.), the Florida Department of Health (Department) submits this annual report for the Rare Disease Advisory Council (Council) to the Governor and the State Surgeon General.

This report describes the activities and findings of the Council during Fiscal Year (FY) 2024-25 and provides ongoing recommendations regarding research and care to improve health outcomes for Floridians affected by rare diseases. Key accomplishments include:

- Implemented the 51-500 Rare Disease Prevalence Data Initiative to identify and compile data for the top 51-500 rare diseases affecting Floridians. These data will provide the groundwork for better understanding the prevalence of rare diseases in Florida and the needs of individuals and families.
- Established grant review guidelines for the Andrew John Anderson Pediatric Rare Disease Research Grant program per section 381.991, F.S.
- Collaborated with rare disease advisory councils in Minnesota, Pennsylvania, and Tennessee to share best practices.
- Continued its work to improve centralized rare disease research resources, maximize its subcommittees and their work, and build partnerships in rare disease care, research, and policy innovation.

Throughout FY 2024-25, the Council worked on recommendations to advance Florida's support for individuals with rare diseases.

- Along with rare disease data collection, the Council recommendations support disaster preparedness for individuals with a rare disease, improvements in health care for rare diseases, strengthening insurance support, greater public awareness, tools to streamline rare disease diagnoses, and building research through a Florida Centers of Excellence for Rare Disease Research and Care.

With this report, the Council affirms its commitment to identifying ways to close diagnostic gaps, support earlier access to care, and foster statewide collaboration through data-informed rare disease strategies and policy.

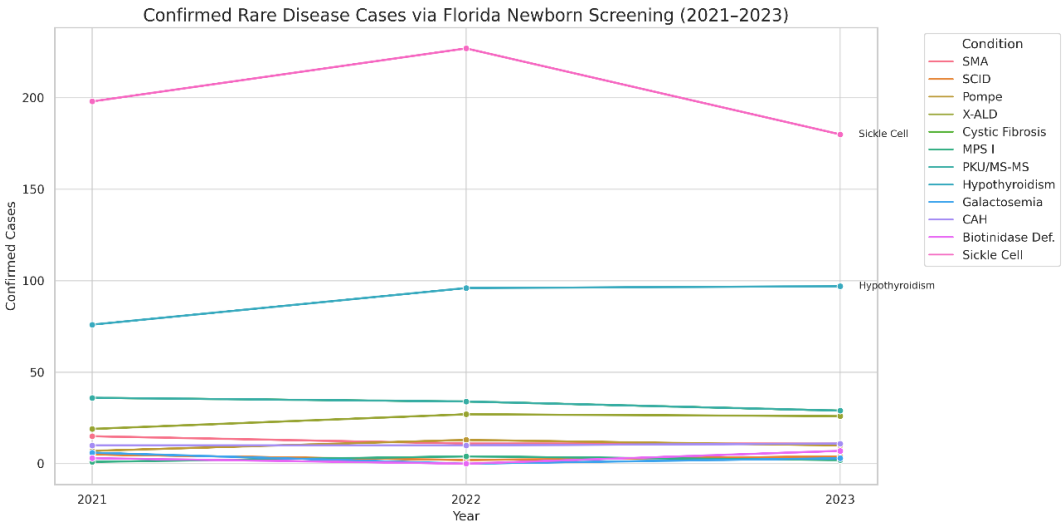
Rare Disease Snapshot

Rare disease, as defined by the Orphan Drug Act, diagnoses directly influence fewer than 200,000 individuals annually in the U.S., meaning these conditions are confirmed in this specific group each year. However, their broader consequences extend to an estimated 25 to 30 million Americans, including roughly 2.2 million Floridians, or 1 in 10 residents, encompassing not only those diagnosed but also families, caregivers, and communities affected by the diseases' social, emotional, or economic burdens. Over 7,000 rare diseases exist, with up to 80% linked to genetic causes, yet only 5% of these diseases have FDA-approved treatments, resulting in significant care gaps.[2]

Nationally, the average time for diagnosis is 4.8 years, with patients often seeing more than seven specialists during their lifetime. Tragically, 30% of affected children die before age 5. The economic burden of rare diseases is significant, affecting 15.5 million Americans with 379 conditions, costing $997 billion in 2019, including $449 billion in medical expenses and $437 billion in lost productivity. This impact rivals the national economic impact of cancer and heart failure.[3]

Newborn screening plays a critical role in identifying a rare disease. Early detection enables timely intervention and care planning, significantly improving outcomes. However, it is important to note that while newborn screening is vital, it captures only a small fraction of the over 7,000 known rare diseases, and children constitute approximately half of those affected by these conditions. Per section 383.14, F.S., the Genetics and Newborn Screening Advisory Council provides newborn screening oversight. Figure 1 shows the number of confirmed cases of rare diseases detected through the Newborn Screening Program between 2021-2024 (additional data are provided in the appendices).

Figure 1: Confirmed Rare Disease Cases through Newborn Screening (2021–24)



Source: Florida Department of Health, Newborn Screening Program (see appendix 1). (2024)

In 2024, Florida demonstrated national leadership in rare disease early detection and patient access. Florida's Newborn Screening Program (Program) was given a rating of "A" by the National Organization for Rare Disorders (NORD).[4] The Program screened more than 221,000 infants and identified over 9,000 with potentially rare conditions. Of those screened, more than 200 infants were confirmed to have rare diseases such as spinal muscular atrophy (SMA), severe combined immunodeficiency (SCID), Pompe disease, and phenylketonuria (PKU), enabling earlier intervention and care planning.

Florida also expanded access to specialized care through participation in the Interstate Medical Licensure Compact (IMLC). Participation in the IMLC can reduce delays in diagnosis and improve patient-provider matching. In parallel, Medicaid coverage for medical nutrition, which is critical to the management of metabolic disorders, was strengthened and helped contribute to the state earning a "B" rating from NORD in this area.[8]

Rare Disease Advisory Council: Overview and Functions

Purpose and Mission

Established in 2021 under section 381.99, F.S., the Council serves as an advisory body to the Governor and State Surgeon General. The Council's goal is to identify strategies that can improve health outcomes for the approximately 2.2 million Floridians (1 in 10) with rare diseases. The Council's mission is to provide insights and make recommendations that may enhance early detection, treatment access, and provider awareness through policy development, research advancement, and care coordination.

Composition

The Council comprises 22 members appointed by the Governor, President of the Senate, and Speaker of the House of Representatives. Members serve 4-year terms and reflect various expertise from state agencies, health care, research, and patient advocacy.

Statutory Responsibilities and Engagement

The Council's duties include consulting with experts, soliciting public input, promoting research, creating provider strategies, and providing input on issues like disaster preparedness. The Council engages through quarterly meetings, subcommittee work, and national collaborations with other Rare Disease Advisory Councils to recommend best practices and align with federal opportunities. An annual report, due by July 1, details activities, findings, and recommendations, and is presented to the Governor and the State Surgeon General. The report is posted on the Department's website.

Long-Term Priorities

The Council's long-term priorities focus on strategies to address:

- Reducing delays in diagnosis and encouraging the use of tools and training that enhance early and accurate diagnoses.
- Understanding any lack of treatment challenges and promoting research and access to therapies for rare diseases.
- Advocating for policies to reduce financial burdens for patients and reduce out-of-pocket expenses.
- Recommending improved access to expertise across Florida and looking for opportunities to encourage more specialists.
- Promoting and educating health care professionals and the public on the burden of rare diseases to increase awareness.
- Supporting patients and families by encouraging enhanced support services for individuals and caregivers.

Council Membership

Members:

- **Gubernatorial Appointees**:
    - Department of Health - Melissa Jordan, MS, MPH, *Chair*
    - Department of Education - Kathyrn Hebda, MM, *Vice Chair*
    - Agency for Health Care Administration - Ann Dalton, MM
    - Office of Insurance Regulation - Alexis Bakofsky
    - Biotechnology - Jonathan Hawayek, MBA
    - Health Insurance - Scott McClelland, PharmD
    - Registered Nurse - Colleen Bartlett, DNP, ARNP, CPNP, FNP-C
    - Pharmacist - Blake Shay, PharmD

- **Senate Appointees**:
    - Academic Researcher - Barry Byrne, MD, PhD
    - Physician - Divya Patel, DO, MBA
    - Self-advocate - India Steinbaugh, MPH
    - Caregivers - Adam Anderson, State Representative; Zana Dupee, JD
    - Care Organization Representative - Anita Davis, PT, DPT, FNCP, CNPT

- **House Appointees**:
    - Academic Researcher - Mustafa Tekin, MD
    - Physician - Rajan Wadhawan, MD, MMM
    - Self-advocate - Jessica O'Reilly, JD
    - Caregivers - Eric Biernacki, JD, LLM; Jennifer Sutherland
    - Care Organization Representative - Rebekah Dorr

Subcommittees:

- **Academic Research Institutions**
    - Chair: Mustafa Tekin, MD
    - Members: Barry Byrne, MD, PhD; Adam Anderson, State Representative; Rajan Wadhawan, MD, MMM; Divya Patel, DO, MBA
- **Health Care Providers**
    - Chair: Anita Davis, PT, DPT, FNCP, CNPT
    - Members: Zana Dupee, JD; India Holroyd, MPH; Jessica O'Reilly, JD; Colleen Bartlett, DNP, ARNP, CPNP, FNP-C; Rebekah Dorr
- **State Agencies**
    - Chair: Melissa Jordan, MS, MPH
    - Members: Ann Dalton, MM; Kathy Hebda, MM; Jonathan Hawayek, MBA; Eric Biernacki, JD, LLM; Blake Shay, PharmD; Scott McClelland, PharmD; Jennifer Sutherland; Alexis Bakofsky

Summary of Council Meetings

July 2024 through June 2025

Meeting Type	Date	Key Discussion Points/Outcomes
Full Council	July 17, 2024	• Reviewed 2024 annual report • Discussed Medicaid whole genome sequencing updates • Proposed new subcommittees (data, grants, resources)
Grant Guidelines Workgroup	Aug. 13, 2024	• Reviewed statutory requirements, national grant standards, and ethical practices • Set timeline for October guideline approval
Health Care Providers Subcommittee	Aug. 14, 2024	• Appointed Anita Davis, PT, DPT as Chair • Reaffirmed focus on education and diagnostic support recommendations
State Agencies Subcommittee	Aug. 15, 2024	• Elected Melissa Jordan, MS, MPH as Chair • Reviewed term limits, initiated statewide agency resource assessment, and discussed disaster preparedness and survey planning
Academic Research Institutions Subcommittee	Aug. 16, 2024	• No quorum: Meeting held but no decisions recorded
Grant Guidelines Workgroup	Sept. 16, 2024	• Approved Conflict of Interest Policy and Grant Review Guidelines • Finalized draft for October 7 Council meeting
Health Care Providers Subcommittee	Sept. 17, 2024	• Reviewed past recommendations and initiated cross-subcommittee survey planning
State Agencies Subcommittee	Sept. 18, 2024	• Outlined interagency survey goals • Discussed integrating emergency preparedness partners
Academic Research Institutions Subcommittee	Sept. 20, 2024	• Elected Mustafa Tekin, MD as Chair, • Reviewed prior minutes • Discussed rare disease research survey and hybrid meeting plans
Full Council	Oct. 7, 2024	• Presented Pediatric Rare Disease Grant recipient updates • Approved grant guidelines and conflict of interest policy
Full Council	Dec. 11, 2024	• Held panel with three other state Rare Disease Advisory Councils on funding, surveys, and special projects • Reviewed grant cycle milestones and application timeline
Health Care Providers Subcommittee	Jan. 9, 2025	• No quorum: Meeting rescheduled
Meeting Type	**Date**	**Key Discussion Points/Outcomes**

State Agencies Subcommittee	Jan. 29, 2025	• Reviewed Medicaid data, rare disease prevalence (discussed Medicaid inclusions, transportation, caregiver support, and tutoring) • Provided updates on insurance barriers and 2025 grant cycle
Full Council	Feb. 20, 2025	• Highlighted Rare Disease Day • Updated Pediatric Rare Disease Grant Program • Discussed workforce support, survey models • Reviewed 51–500 rare disease prevalence data
Academic Research Institutions Subcommittee	March 24, 2025	• Identified need for centralized rare disease research directory • Set goals to map research activity, • Proposed hybrid model for voluntary submissions and outreach
Health Care Providers Subcommittee	March 25, 2025	• No quorum: Meeting held but no decisions recorded
State Agencies Subcommittee	April 8, 2025	• Reviewed past recommendations • Identified gaps and targets for 2025 refinements
Academic Research Institutions Subcommittee	April 9, 2025	• Finalized recommendations for centralized research directory and rare disease designation for institutions
Health Care Providers Subcommittee	April 16, 2025	• No quorum: Meeting held but no decisions recorded
Full Council	April 23, 2025	• Reviewed recommendations • Heard subcommittee reports • Recommended funding for Pediatric Rare Disease Grant Program

- All meetings were announced in Florida Administrative Register and on the Department website with virtual/teleconferencing options.
- Subcommittees met regularly with quorum achieved unless noted.
- The Grant Guidelines Workgroup was a temporary working group.

Key Accomplishments (FY 2024-25)

Over the past year, the Council, through subcommittee efforts and cross-sector partnerships, has recommended strategies and fostered collaborations to strengthen Florida's rare disease infrastructure.

Key achievements supported by the Council include:

- Reviewing the Andrew John Anderson Pediatric Rare Disease Grant Program applications, which provides $500,000 annually to support innovative clinical research. The Council recommended guidelines and a conflict-of-interest policy to use when prioritizing funding for the Andrew John Anderson Pediatric Rare Disease Research Grant.
- Facilitated an interstate council collaboration effort where they engaged with Minnesota, Pennsylvania, and Tennessee Rare Disease Advisory Councils to share best practices on data usage, surveys, and funding models, to inform Florida's strategic recommendations.
- Continuing its earlier work to identify the top 50 rare diseases, the council supported the compilation and analysis of the top 51-500 rare diseases impacting Floridians. When complete, the analysis of these data, based on International Classification of Diseases, Tenth Revision (ICD-10), and National Drug Codes (NDC) data, will provide a foundation for the consideration of a statewide rare disease snapshot.
- Conducted an academic research mapping assessment to identify gaps in centralized research resources and recommend a hybrid model to promote collaboration among Florida's academic institutions.
- Worked to construct a framework for delegating tasks to subcommittees, leveraging external partners (e.g., National Institutes of Health, NORD) and developing low-burden tools (e.g., online surveys, FLHealthCHARTS) to advance priorities.

These efforts demonstrate Florida's growing capacity for rare disease care, research, and policy innovation.

Council Recommendations for FY 2025-26

The Council presents the following recommendations to guide stakeholders in strengthening Florida's rare disease framework and to consolidate subcommittee priorities to ensure comprehensive, non-redundant strategies.

1. **Recommend Data Collection to Understand Florida's Rare Disease Population:** Encourage the compilation of prevalence and incidence data using ICD-10 and national datasets to develop a Florida-specific rare disease catalog, enabling comparisons with other states for policy and resource allocation.
2. **Promote Disaster Preparedness for Individuals Living with a Rare Disease**: Recommend assessing needs and creating educational materials to support patients and emergency staff, leveraging resources like Florida's Special Needs Shelter Registry.
3. **Encourage Enhanced Health Care Services for Individuals Living with a Rare Disease:** Recommend that the Florida Department of Health, Agency for Health Care Administration, and Department of Education enhance services through data sharing, needs assessments, and staff training.
4. **Support Improved Insurance Support for Individuals Living with a Rare Disease:** Recommend the Florida Office of Insurance Regulation (OIR) strengthen support by identifying commercial health plan data, ensuring the promotion of fair and consistent benefit access, and training of staff.
5. **Promote Provider Education and Awareness for Rare Diseases:** Encourage enhanced provider education through awareness campaigns, specialist identification, and interdisciplinary care team support to improve diagnostic accuracy and patient outcomes.
6. **Encourage Streamlined Rare Disease Diagnosis:** Recommend developing tools like symptom checklists, decision trees, and infographics to guide providers and reduce diagnostic delays.
7. **Foster Collaboration and Funding for Rare Disease Research:** Recommend strengthening research through partnerships with stakeholder groups including the NORD and targeting sustained funding for programs like the Pediatric Rare Disease Grant Program.
8. **Encourage Centers of Excellence for Rare Disease Research and Care:** Recommend developing a framework for rare disease Centers of Excellence to enhance research, clinical trials, and patient care, including mapping institutions and promoting genome sequencing.

Appendix

Appendix 1: Rare Diseases Identified by Florida's Newborn Screening Program (2021-24)

Florida's Newborn Screening Program screens over 99% of live births and plays a critical role in the early identification of rare conditions. Many of these diseases, such as SMA, SCID, Pompe Disease, and X-ALD, require early intervention to prevent severe disability or death.

The chart below displays the number of confirmed cases for a selection of these conditions over the past four years, underscoring the value of newborn screening in the rare disease landscape.[5]

Condition	2021	2022	2023	2024*	% Change (2021-23)
Spinal Muscular Atrophy (SMA)	15	11	11	3	--26.7
Severe Combined Immunodeficiency (SCID)	5	2	4	0	-20.00
Pompe Disease	7	13	10	6	42.86
X-Linked Adrenoleukodystrophy (X-ALD)	19	27	26	10	36.84
Cystic Fibrosis	36	34	29	13	-19.44
Mucopolysaccharidosis Type I (MPS I)	1	4	2	2	100.00
Phenylketonuria (PKU)/MS-MS	36	34	29	17	-19.44
Congenital Hypothyroidism	76	96	97	93	27.63
Galactosemia	6	0	3	1	-50.00
Congenital Adrenal Hyperplasia (CAH)	10	10	11	1	10
Biotinidase Deficiency	3	0	7	3	133.33
Sickle Cell Disease	198	227	180	29	-9.1

Source: Florida Department of Health's Newborn Screening Program (2024). Confirmed Rare Disease Cases via Newborn Screening, 2021-24.

**2024 represents only partial year data.*

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[1] Orphan Drug Act, H.R.5238, 97th Congress (1983); Global Genes, Rare Disease Impact Report (2024); U.S. Department of Health and Human Services, FAQs About Rare Diseases, Genetic and Rare Diseases Information Center (2021).
[2] National Organization for Rare Disorders (NORD), Rare Disease Overview (2024); U.S. Food and Drug Administration (FDA), Rare Diseases at FDA (2024).
[3] Global Genes. (2024). "Rare Disease Impact Report." Retrieved from globalgenes.org, Yang, G., et al. (2022). "The national economic burden of rare disease in the United States in 2019." Orphanet Journal of Rare Diseases, 17, 163. https://pubmed.ncbi.nlm.nih.gov/35414039/
[4] NORD. (2024). *State Report Card 2024*. Retrieved from rarediseases.org.
[5] Florida Department of Health, Newborn Screening Program. (2024). Confirmed Rare Disease Cases via Newborn Screening, 2021–2024. Unpublished data provided to the Florida Rare Disease Advisory Council.