RARE DISEASE ADVISORY COUNCIL

Fiscal Year 2025-26 Annual Report

July 1, 2026

Ron DeSantis
Governor

Joseph A. Ladapo, MD, PhD
State Surgeon General

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Executive Summary

Rare diseases are defined by the federal Orphan Drug Act of 1983 as conditions affecting fewer than 200,000 individuals in the United States each year. Although individually uncommon, they collectively affect an estimated 25 to 30 million Americans, including approximately 2.2 million Floridians—about one in 10 residents. More than 7,000 rare diseases are known, up to 80% have a genetic basis, and only about 5% currently have a U.S. Food and Drug Administration approved treatment. As a result, individuals suffering rare disease symptoms often experience extended diagnostic delays—averaging nearly five years—and complex care needs. The national economic burden of rare diseases is substantial, accounting for an estimated $997 billion in medical costs and lost productivity in 2019. Children are disproportionately burdened by rare diseases, and approximately 30% of children diagnosed with a rare disease die before age five.[1]

Newborn screening serves as a critical tool for early detection of certain rare conditions, helping enable timely interventions. However, screening identifies only a small portion of the more than 7,000 recognized rare diseases. In Florida, oversight of newborn screening is provided by the Genetics and Newborn Screening Advisory Council under section 383.14, Florida Statutes.

Pursuant to section 381.99, F.S., the Florida Department of Health submits this annual report summarizing the activities and recommendations of the Rare Disease Advisory Council (Council) for Fiscal Year (FY) 2025–26. The Council's work advances understanding of Florida's rare disease population and strengthens the state's capacity to support individuals with a rare disease diagnosis.

During FY 2025–26, the Council achieved several important accomplishments that address data quality, provider awareness, and care coordination statewide. These accomplishments include:

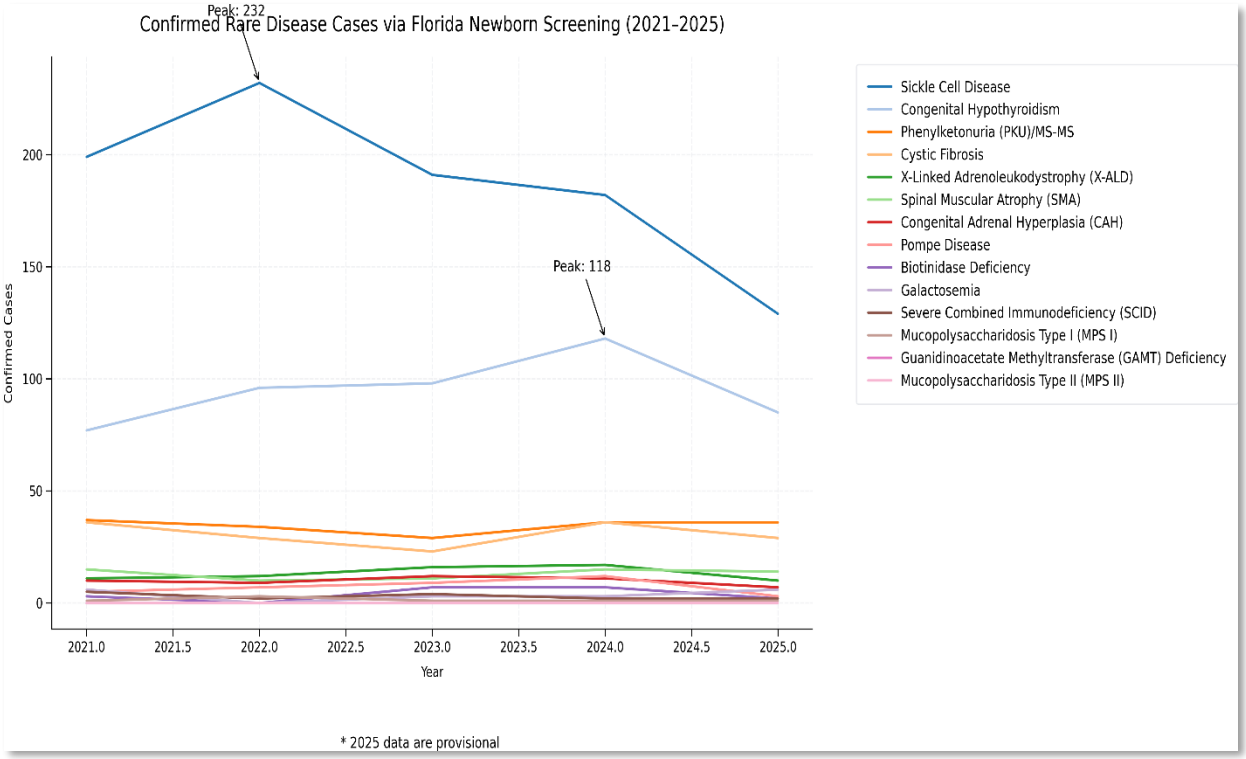
- Continuing collection of rare disease data from the Office of Insurance Regulation and cross-referencing findings with Medicaid data to better estimate impacted populations.
- Updating Medicaid utilization data to reflect current service use trends among individuals with rare diseases.
- Reviewing Medicaid nonemergency transportation and lodging supports to help families access specialized care.
- Identifying evidence to develop a "red flag" clinical indicator tool intended to support earlier rare disease recognition by providers.
- Mapping Chronic Complex Clinics across Florida to improve disaster preparedness, care coordination, and sharing of best practices among pediatric and adult providers.

The Council's ongoing activities emphasize integrated data collection, provider education, improved diagnostic pathways, and strengthened collaboration across institutions. These priorities aim to enhance early detection, expand access to specialized care, and ultimately improve health outcomes for Floridians living with rare diseases.

Rare Disease Snapshot

Florida's commitment to early detection and access to care for individuals with rare diseases is supported by the strong performance in newborn screening. The Florida Newborn Screening Program received an "A" rating from the National Organization for Rare Disorders (NORD), reflecting robust screening policies and alignment with recommended conditions. Florida screens approximately 250,000 newborns annually for roughly 60 rare conditions including spinal muscular atrophy (SMA), severe combined immunodeficiency (SCID), Pompe disease, and phenylketonuria (PKU)—supporting early diagnosis and timely care planning.[2]

Figure 1: Confirmed Rare Disease Cases through Newborn Screening (2021-25)



Source: Florida Department of Health, Newborn Screening Program (see appendix 1). (2025)

In addition, since 2024 when Florida joined the Interstate Medical Licensure Compact (IMLC),[3] access to specialized medical providers has expanded, helping reduce delays in diagnosis and earning the state a Pass rating from the National Organization for Rare Disorders (NORD).

Rare Disease Advisory Council: Overview and Functions

Purpose and Mission

Established in 2021 in section 381.99, F.S., the Council serves as an advisory body to the Governor and State Surgeon General. The Council's purpose 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 (RDACs) to recommend best practices that align with federal guidelines. 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 of Health's website.

Long-Term Priorities

The Council's long-term priorities focus on six strategies.

- 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

- **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
- **President of the Senate Appointees:**
  - Academic Researcher - Barry Byrne, MD, PhD
  - Physician - Divya Patel, DO, MBA
  - Self-advocate - India Holroyd, MPH
  - Caregivers - Adam Anderson, State Representative; Zana Dupee, JD
  - Care Organization Representative - Anita Davis, PT, DPT, FNCP, CNPT
- **Speaker of the 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
- **Vacant Council Positions:**
  - Geneticist
  - Hospital administrator from a hospital providing care to rare disease patients

Council 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, Eric Biernacki, JD, LLM: Kathyrn Hebda, MM: Melissa Jordan, MS, MPH
- **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, Ann Dalton, MM; Jonathan Hawayek, MBA; Blake Shay, PharmD; Scott McClelland, PharmD; Jennifer Sutherland; Alexis Bakofsky

Summary of Council Meetings

July 2025 through June 2026

Meeting Type	Date	Key Discussion Points/Outcomes

State Health Providers	Aug 14, 2025	• Two priorities set for FY 2024–25:
		  ○ Resource assessment and rare-disease survey
		  ○ Collaborate with RDACs on disaster preparedness

Full Council	Aug 27, 2025	• Support creation of centralized rare disease hub • Approved restructuring to resolve quorum issues • Affirmed goals for expanded newborn sequencing • Instructed members to review and renew terms
Full Council	Nov 12, 2025	• Agreed to pursue a centralized rare disease resource website • Develop criteria for Centers of Excellence • Confirmed ongoing work on registry, data analysis, and preparedness • Agreed to collaborate on mapping centers and developing provider education tools
Full Council	Jan 15, 2026	• Received research progress updates from Florida State University and University of Miami grant projects • Notified council members of Senate Bill 1060 establishing a Neurofibromatosis Grant Program under Council oversight • Flagged additional newborn screening and rare disease policy proposals for awareness and discussion • Encouraged sharing Rare Disease Day events and noted Chair Jordan's SB 1060 presentation on the Florida Channel

Full Council	March 4, 2026	• Update on Barth Syndrome research from University of Miami grant project
		• Subcommittee updates
		  ○ Health Care Providers – Reviewed strategies for identifying rare disease populations and discussed barriers such as transportation, disaster preparedness, and diagnostic tools.
		  ○ Academic Research Institutions – Discussed creating a statewide rare disease resource website and exploring a Centers of Excellence network.
		• Member updates – Shared Rare Disease Day activities, key legislative items, proposed programs, newborn screening additions, and questions about the recent grant Letter of Intent cycle.

Grant Workgroup	Apr 29, 2026	Andrew John Anderson Pediatric Grant Awards • Identified top proposals • Approved funding of two projects • Next steps outlined
Full Council	June 18, 2026	Upcoming

- All meetings were announced in Florida Administrative Register and on the Department of Health's website with virtual/teleconferencing options.
- Listed is the final 2025–26 subcommittee meeting, held before subcommittees were integrated into breakout sessions during full council meetings.
- The grant work group gathers periodically to assess proposals and approve funding allocations.

Key Accomplishments (FY 2025-26)

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:

- Continued collection of data from the Office of Insurance Regulation to capture the size of the population with rare disease based on coding and utilization. Data to be cross referenced with Medicaid information.

- Medicaid data has been updated to reflect utilization.

- Reviewed Medicaid's non-emergency transportation policy and possible lodging support to assist families with rare disease in accessing specialty care.

- Identified articles to guide the development of a red flag indicator tool rather than screening tool. Articles under review to determine next steps for utilization.

- Mapping Chronic Complex Clinics across the state that serve adults and children.
- Efforts to coordinate resources, especially related to disaster preparedness and sharing best practices.

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

Council Recommendations for FY 2026-27

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. **Strengthen Statewide Rare Disease Data Collection**
   Continue to update and merge data from major insurance carriers, including Medicaid and state employee health plans.

2. **Enhance Disaster Preparedness for Individuals with Rare Diseases**
   Continue developing partnerships with Chronic Complex Clinics to share best practices for locating and transporting individuals with special needs to ensure their inclusion and safety during emergencies.

3. **Expand Provider Education and Awareness**
   Encourage enhanced provider education through awareness campaigns, specialist identification, and interdisciplinary care team support to improve diagnostic accuracy and patient outcomes.

4. **Improve Diagnostic Pathways for Rare Diseases**
   Review and develop educational resources to help providers better recognize the possibility of rare diseases in their patient populations.

5. **Establish a Statewide Collaboration and Information-Sharing Framework**
   Develop a formal process for collaboration between institutions and providers, including a shared structure for exchanging data and research to prevent duplication of efforts, and creating a centralized rare disease resource website for Florida to support unified access to information.

Appendix 1

Appendix 1: Rare Diseases Identified by Florida's Newborn Screening Program (2021-25)

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 Spinal Muscular Atrophy, Severe Combined Immunodeficiency, Pompe Disease, and X-Linked Adrenoleukodystrophy, 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 five years, underscoring the value of newborn screening in the rare disease landscape.[5]

Newborn Screening Data 2021-2025					
Condition	2021	2022	2023	2024	2025*
Spinal Muscular Atrophy (SMA)	15	10	11	15	14
Severe Combined Immunodeficiency (SCID)	5	2	4	2	2
Pompe Disease	5	7	9	12	3
X-Linked Adrenoleukodystrophy (X-ALD)	11	12	16	17	10
Cystic Fibrosis	36	29	23	36	29
Mucopolysaccharidosis Type I (MPS I)	1	3	1	1	1
Congenital Hypothyroidism	77	96	98	118	85
Phenylketonuria (PKU)/MS-MS	37	34	29	36	36
Galactosemia	6	0	3	3	6
Congenital Adrenal Hyperplasia (CAH)	10	9	12	11	7
Biotinidase Deficiency	3	0	7	7	2
Sickle Cell Disease	199	232	191	182	129
Mucopolysaccharidosis Type II (MPS II)				0	0
Guanidinoacetate Methyltransferase (GAMT) Deficiency					0

*This data is provisional. 2025 represents only partial year data.

Source: Florida Department of Health's Newborn Screening Program (2025). Confirmed Rare Disease Cases via Newborn Screening, 2021-25.

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[1] Orphan Drug Act, H.R.5238, 97th Congress (1983); Global Genes, Rare Disease Impact Report (2025); 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 (2025); U.S. Food and Drug Administration (FDA), Rare Diseases at FDA (2025).

[3] National Organization for Rare Disorders (NORD), State Report Card (2025), Member of Interstate Medical Licensure Compact (IMLC). Retrieved from raredisease.org.

[4] Florida Department of Health, Newborn Screening Program. (2025). Confirmed Rare Disease Cases via Newborn Screening, 2021–2025. Unpublished data provided to the Florida Rare Disease Advisory Council.