Rare Disease Advisory Council

Legislative Report

Fiscal Year 2022/2023

July 1, 2023

Ron DeSantis

Governor

Joseph A. Ladapo, MD, PhD

State Surgeon General

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Executive Summary

The Florida Department of Health (Department), Division of Community Health Promotion is pleased to present the second annual legislative report of the Rare Disease Advisory Council (RDAC) in accordance with section 381.99, Florida Statutes.

In June 2021, 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. Council members hold a shared vision: To improve health outcomes for individuals residing in Florida who have rare diseases.

During the initial planning year, the council developed meaningful recommendations and objectives directed to improve the lives of people with rare diseases in Florida. During this past year, the council began to work toward meeting goals identified for year one. The information gathered has secured the groundwork for identifying what the state of Florida is currently doing to serve individuals with rare diseases, what services are currently in place, and Florida's current protocols for diagnosing individuals with rare diseases.

July 1, 2023

The Rare Disease Advisory Council was established in section 381.99, Florida Statutes, to assist the Department of Health (Department) in providing recommendations to improve health outcomes for individuals residing in the state who have a rare disease.

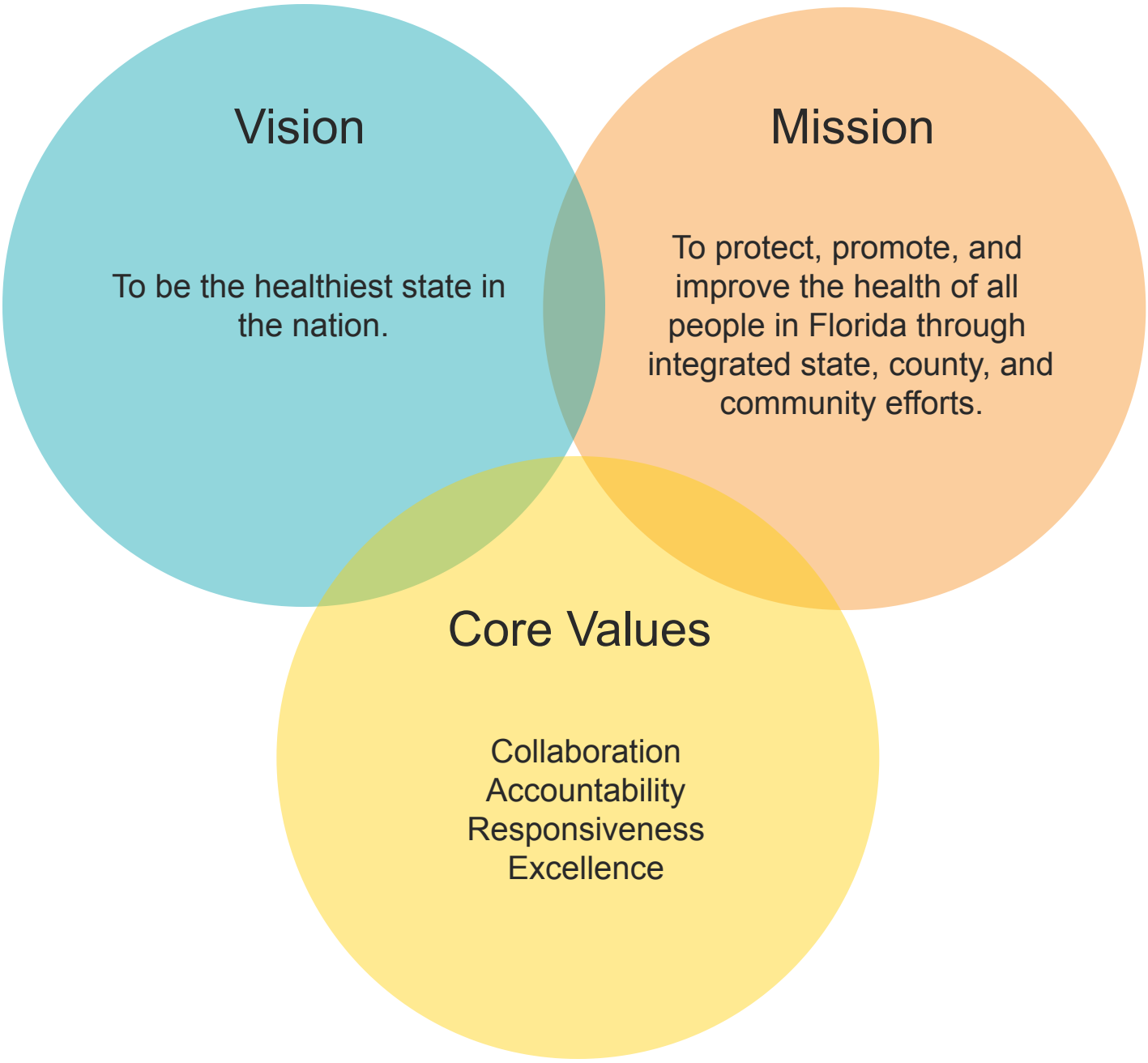
On behalf of the members of the Florida Rare Disease Advisory Council (RDAC), it is my pleasure to submit the second annual legislative report as outlined in section 381.99, Florida Statutes. This report documents the collaborative efforts of the RDAC over the past year and outlines their dedication to developing recommendations to improve 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 RDAC 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 RDAC seeks to encourage research on rare diseases in Florida.

Sincerely,

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

Vision, Mission, and Core Values



RDAC Membership

An individual who is, or was previously, a caregiver for individuals with a rare disease:
Eric Biernacki
Caregiver
Appointed by Speaker of the House of Representatives

A physician who is licensed under Chapter 458 or Chapter 459, Florida Statutes, and practicing in this state with experience in treating rare diseases:
Barry Byrne, MD, PhD
Director, Powell Gene Therapy Center
Associate Chair, Department of Pediatrics
Professor, Pediatrics and Molecular Genetics & Microbiology
University of Florida
Appointed by the President of the Senate

A representative of the Agency for Health Care Administration:
Ann Dalton, MM
Bureau Chief, Bureau of Medicaid Policy
Florida Agency for Health Care Administration
Appointed by the Governor

A representative of organizations in this state which provide care or other support to individuals with rare diseases:
Anita Davis, PT, DPT, FNCP, CNPT
Physical Therapist
Brooks Rehabilitation
Appointed by the President of the Senate

A representative of organizations in this state which provide care or other support to individuals with rare diseases:
Rebekah Dorr
Director of Clinical Patient Advocacy
Myasthenia Gravis Hope Foundation
Appointed by the Speaker of the House of Representatives

An individual who is, or was previously, a caregiver for individuals with a rare disease:
Zana Dupee, JD
Caregiver
Appointed by the President of the Senate

RDAC Membership

A representative of the biotechnology industry:
Jonathan Hawayek, MBA
Head of State Government Affairs
Spark Therapeutics, Inc.
Appointed by the Governor

A representative of the Department of Education:
Kathryn Hebda, MM - Council Vice Chair
Chancellor, Florida College System
Florida Department of Education
Appointed by the Governor

A representative of the Department of Health:
Melissa Jordan, MS, MPH - RDAC Chair
Assistant Deputy Secretary for Health
Florida Department of Health
Appointed by the Governor

A representative of health insurance companies:
Scott McClelland, PharmD
Vice President, Commercial and
Specialty Pharmacy Programs and
Health Care Solutions
Florida Blue
Appointed by the Governor

An individual who is 18 years of age or older who has a rare disease:
Jessica O'Reilly, JD
Self-Advocate
Appointed by the Speaker of the House of Representatives

A representative from an academic research institution in Florida, which receives grant funding for research regarding rare diseases:
Divya Patel, DO, MBA
Program Director, Interstitial Lung Disease
Program Director, Sarcoidosis
Associate Professor of Medicine
University of Florida
Appointed by the President of the Senate

RDAC Membership

An individual who is 18 years of age or older who has a rare disease:
India Steinbaugh, MPH
Self-Advocate
Appointed by the President of the Senate

An individual who is, or was previously, a caregiver for individuals with a rare disease:
Jennifer Sutherland
Caregiver
Appointed by the Speaker of the House of Representatives

A representative from an academic research institution in this state which receives grant funding for research regarding rare diseases:
Mustafa Tekin, MD
Division Director, Clinical and Translational Genetics
University of Miami
Appointed by the Speaker of the House of Representatives

A physician who is licensed under Chapter 458 or Chapter 459, Florida Statutes, and practicing in this state with experience in treating rare diseases:
Rajan Wadhawan, MD, MMM
Senior Executive Officer, AdventHealth
for Children and AdventHealth for Women
Appointed by the Speaker of the House of Representatives

An individual who is, or was previously, a caregiver for individuals with a rare disease:
Vacant

A representative of the Office of Insurance Regulation:
Vacant

A pharmacist who is licensed and practicing in this state who has experience with drugs that are used in the treatment of rare diseases:
Vacant

One geneticist practicing in this state:
Vacant

RDAC Membership

One registered nurse or advanced practice registered nurse who is licensed and practicing in this state with experience in treating rare diseases:
Vacant

One hospital administrator from a hospital in this state which provides care to individuals diagnosed with rare diseases:
Vacant

Introduction

The Orphan Drug Act defines a rare disease as a disease or condition that affects fewer than 200,000 individuals in the United States.[1] According to the National Institutes of Health, the total number of Americans living with rare diseases is estimated to be between 25 to 30 million individuals, or approximately 10 percent of the total national population.[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.

Rare diseases include genetic disorders, infectious diseases, cancers, and other various pediatric and adult conditions.[2] 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.[3] 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.[4] There are a number of 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.[5] 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. 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.[6] 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.

1) H.R.5238 - 97th Congress (1981-1982): Orphan Drug Act. (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) Rare Genetic Diseases. Genome.gov. (2018, April 13). Retrieved from https://www.genome.gov/dna-day/15-ways/rare-genetic-diseases
4) U.S. National Library of Medicine. (n.d.). What is a gene variant and how do variants occur? MedlinePlus Genetics. MedlinePlus. Retrieved from https://medlineplus.gov/genetics/understanding/mutationsanddisorders/genemutation/?msclkid=ba654498cfdf11ec9fb750b3c3c476aa
5) 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.
6) 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-consumers-and-patients-drugs/orphan-products-hope-people-rare-diseases

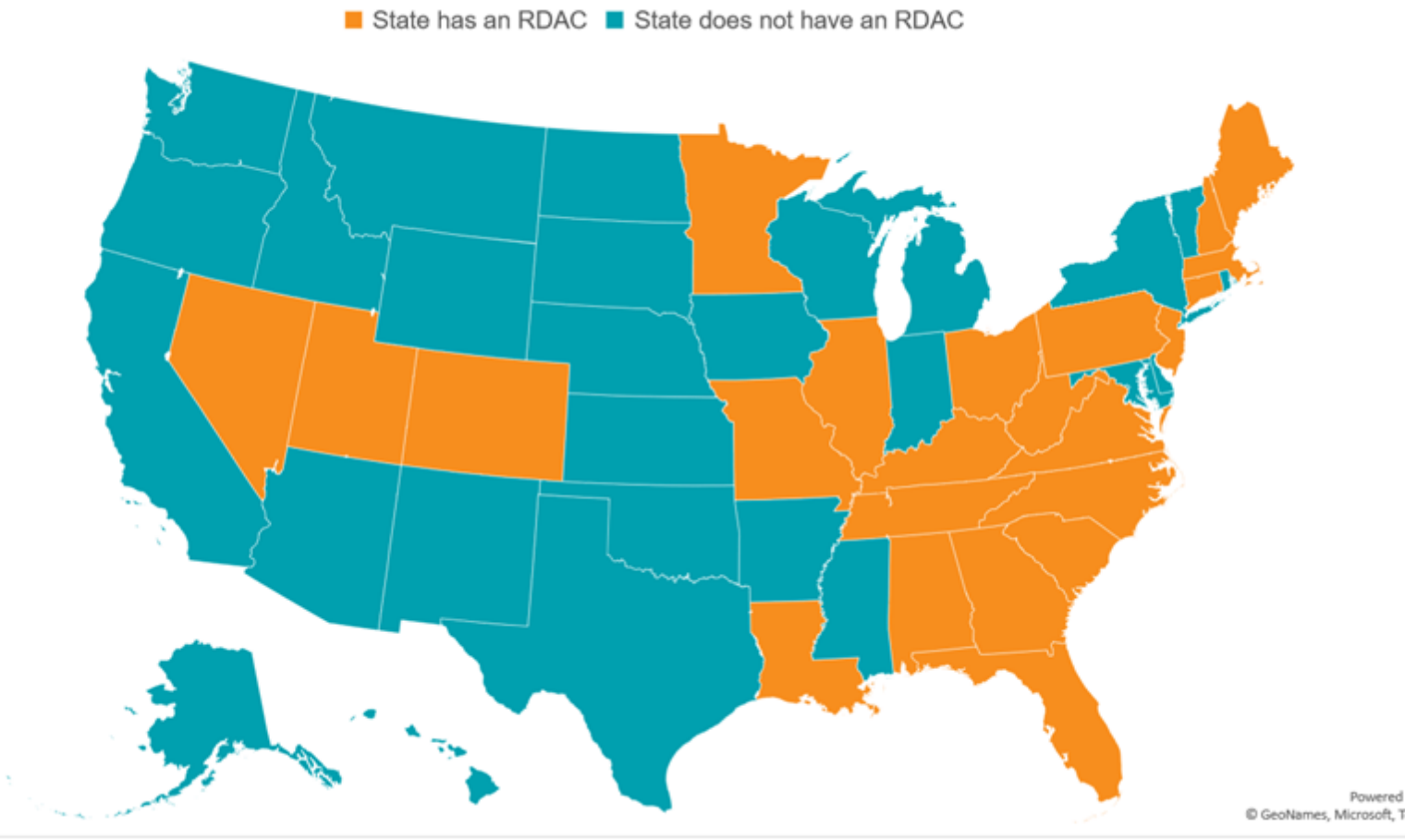
Introduction

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.

Some critical issues the rare disease community faces are the need to increase awareness, diagnostic tools, and access to affordable treatment and cures. Since the inception of the RDAC in 2021, the council has examined those concerns. The RDAC executed year one recommendations and objectives it developed within the planning period. This report provides status updates regarding the accomplishments of the RDAC's work toward meeting year one goals. These status updates also include a summary of the work performed and solutions developed for challenges that were encountered.

Background

On June 21, 2021, Senate Bill 272 was signed into law by Governor Ron DeSantis to establish section 381.99, Florida Statutes — Rare Disease Advisory Council. Florida became the 19th state to pass legislation creating an RDAC. Twenty-three states have established councils to help better represent the rare disease community.



7) *RDACs by State*. NORD (National Organization for Rare Disorders). (2023, March 27). Retrieved from https://rarediseases.org/policy-issues/rare-disease-advisory-councils/.

Functions of the RDAC

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

Consult with experts on rare diseases and solicit public comment to assist in developing recommendations on improving the treatment of rare diseases in this state.

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

Develop recommended strategies for health care providers to be informed on how to more effectively recognize and diagnose rare diseases in order to effectively treat patients. 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.

Fiscal Year 2022-2023 Timeline

August 2022

- The RDAC convened on August 24.

September 2022

- The RDAC convened on September 28.
- The Academic Research Institutions Subcommittee convened on September 26.
- The State Agencies Subcommittee convened on September 27.
- The Health Care Providers Subcommittee Convened on September 27.

October 2022

- The RDAC convened on October 26.
- Year 1 objectives for each subcommittee implementation plan was finalized.
- The RDAC and Subcommittee meetings schedule developed for 2022-2023.

November 2022

- The Academic Research Institution Subcommittee convened on November 22.
- The State Agencies Subcommittee convened on November 30.
- The Health Care Providers Subcommittee convened on November 30.

December 2022

- The Academic Research Institution Subcommittee convened on December 19.
- The State Agencies Subcommittee convened on December 14.
- The Health Care Providers Subcommittee convened on December 14.

January 2023

- The Academic Research Institution Subcommittee convened on January 26.
- The State Agencies Subcommittee convened on January 17.
- The Health Care Provider Subcommittee convened on January 24.

February 2023

- The Academic Research Institution Subcommittee convened on February 23.
- The State Agencies Subcommittee convened on February 21.
- The Health Care Providers Subcommittee convened on February 24.

March 2023

- The Academic Research Institution Subcommittee convened on March 21.
- The State Agencies Subcommittee convened on March 22.
- The Health Care Subcommittee convened on March 16.

April 2023

- The RDAC convened on April 17.

May 2023

- The RDAC convened May 8.

Fiscal Year 2022-2023 Accomplishments

Council Meeting Operations and Procedures

Since July 2022, the full RDAC has convened six times.

Continuation of Subcommittees

The RDAC formed three subcommittees: the Academic Research Institutions Subcommittee, the Health Care Providers Subcommittee, and the State Agencies Subcommittee in order to address the requirements under section 381.99 (4)(b), (4)(c), and (4)(d), Florida Statutes (Appendix A). Since their formation, the Academic Research Institutions Subcommittee, the Health Care Providers Subcommittee, and the State Agencies Subcommittee continue to meet monthly, except during the summer months.

Consultation with Experts on Rare Diseases

Council members have consulted with national and state experts on rare diseases during their efforts to meet year one goals.

Solicitation of Public Comment

Each meeting convened by the RDAC and its subcommittees is publicly posted on the Florida Administrative Register as well as a webpage hosted by the Department for RDAC meeting activities. An invitation for public comment is provided prior to the adjournment of each RDAC meeting.

Recommended Strategies for Academic Research Institutions

Recommendation 1: Support research institutions in Florida.

Year 1 Objectives

Objective 1.1: Identify the components that constitute a best practice “rare disease research institution."

Status: In Progress

Summary: The National Organization for Rare Diseases (NORD) celebrates 40 years of advocacy for those diagnosed with a rare disease. NORD is the leading organization for research, education, and policy change surrounding rare diseases, making it a trusted resource for RDAC. Using the description of the NORD Rare Disease Centers of Excellence (NORD RD CoE) Program (Appendix B), RDAC members have developed a framework for identifying Academic Rare Disease Institutions (Academic RDRI) and Clinical Rare Disease Institutions (Clinical RDRI) within Florida (Appendix C). Entering year two, the RDAC will develop a recommendation and the organization will make these institution designations.

Objective 1.2: Elicit feedback on developed components for a best practice "rare disease research institution."

Status: In Progress

Summary: The Academic Research Institutions Subcommittee continues to expand a list of rare disease providers (Appendix D) and investigators (Appendix E) that will be utilized to elicit feedback on the developed framework for the designation of Rare Disease Research Institutions in Florida. A complete review of the feedback and final decisions on the RDAC components for best practice criteria will be moved to year two.

Recommended Strategies for Academic Research Institutions

Recommendation 2: Promote availability of research institutions to individuals with rare diseases.

Year 1 Objective

Objective 2.1: Promote expanded access to genome sequencing for all individuals with rare diseases.

Status: In Progress

Summary: The Academic Research Institutions Subcommittee plans to continue discussions with members of the State Agencies Subcommittee to identify financial challenges to families needing access to genetic testing for diagnosis and treatment.

Recommended Strategies for Academic Research Institutions

Recommendation 3: Collaborate with state and national experts on complex rare disease cases.

Year 1 Objective

Objective 3.1: Identify state and national rare disease experts available to consult with health care providers on complex rare disease cases.

Status: In Progress

Summary: Academic Research Institutions Subcommittee members generated a draft list of board-certified geneticists (Appendix E) in Florida. Moving forward, these providers will be contacted with an invitation to collaborate with other providers in the development of care plans and treatment of patients diagnosed with a rare disease. Additional providers will be identified based on their respective areas of expertise in year two.

Recommended Strategies for Health Care Providers

Recommendation 1: Promote education on rare diseases to health care providers.

Year 1 Objectives

Objective 1.1: Identify existing educational resources, including continuing education courses for health care providers on various topics related to rare diseases.

Status: Ongoing

Summary: The RDAC aims to be a trusted resource for health care providers in Florida. Health Care Providers Subcommittee members created a list of educational resources health care providers can use when searching for reliable information regarding rare diseases (Appendix F). It is anticipated that this list will continue to grow.

Objective 1.2: Identify educational resources on patient-centered care for health care providers.

Status: Ongoing

Summary: The Health Care Providers Subcommittee developed a list of online educational resources regarding patient-centered care that are available online (Appendix G). Patient and family engagement can improve treatment outcomes and quality of life. These resources will assist providers improved communication techniques to better engage patients and their families in treatment options and through development of the plan of care. It is anticipated that this list will continue to be expanded.

Objective 1.3: Identify educational resources on holistic health care practices for health care providers.

Status: In Progress

Summary: This resource list (Appendix H) aims to assist providers in identifying the relationships between organic or functional elements that affect their patient's individual wellbeing. Holistic approaches include, but are not limited to, acupuncture, biofeedback, massage therapy, chiropractic therapy, and guided imagery. It was proposed to the State Agencies Subcommittee that a questionnaire should be developed and sent to a sampling of patients with rare diseases and their physicians to collect information that can shape the outcome of this objective. This objective will be further examined in year two.

Recommended Strategies for Health Care Providers

Recommendation 1: Promote education on rare diseases to health care providers.

Year 1 Objectives

Objective 1.4: Centralize educational resources on a publicly available website.

Status: In Progress

Summary: This website will serve as a centralized location with resources for patients diagnosed with a rare disease, their families, and their health care team. This remains in a planning phase. The Health Care Providers Subcommittee is researching possible resources to post on the website.

Recommended Strategies for Health Care Providers

Recommendation 2: Promote advancements in the process of achieving a diagnosis for rare diseases.

Year 1 Objective

Objective 2.1: Identify screening tools for health care providers on rare diseases.

Status: In Progress

Summary: Through exploration of existing screening tools, the Health Care Providers Subcommittee has identified there are currently no standard protocols for physicians in the diagnosis of autoimmune diseases. This adds to the difficulty of diagnosis because autoimmune diseases are not only specific, but complicated. Lab testing is not always reliable due to variables. A positive antinuclear antibodies test, for example, may be a sign of an autoimmune disorder but does not provide a clear diagnosis. The physician is responsible for deciding which additional assessments and testing to perform that may lead to a diagnosis, though there are no clear guidelines or protocols. The physician relies on experience and expertise. There are, however, protocols for genetic testing. This includes the Florida Newborn Screening Program. Genetic testing provides a conclusion based on verifiable genetic markers for diagnosis. The Health Care Providers Subcommittee plans to consult with the Academic Research Institutions Subcommittee on this objective to identify centers of excellence for rare diseases instead of developing individual screening plans. Further development of this objective will continue in year two.

Recommended Strategies for Health Care Providers

Recommendation 3: Promote advancements in the process of achieving treatments for rare diseases.

Year 1 Objectives

Objective 3.1: Identify current medical practices and specialists who accept both pediatric and adult individuals with rare diseases.

Status: In Progress

Summary: There is a challenge for patients diagnosed with a rare disease as a child to find a provider that will assume their care once they transition to adulthood. There are even fewer providers that will accept both pediatric and adult populations with rare diseases. The Health Care Providers Subcommittee was able to locate two providers (Appendix I) that accept pediatric and adult patients with rare diseases. The Health Care Providers Subcommittee plans to survey providers in year two.

Objective 3.2: Identify barriers and challenges to medical practices and specialists accepting both pediatric and adult individuals with rare diseases.

Status: Completed

Summary: The Health Care Providers Subcommittee has developed a list of potential barriers and challenges faced by medical practices and specialists to accepting pediatric and adult individuals with rare diseases. This includes the following reasons: 1) the practice does not accept the individual's insurance, 2) the insurance carrier does not contract with the provider, 3) the providers in the practice are not trained in treating one group or the other. A provider survey may assist with identifying additional factors that may affect the ability of a practice to accept pediatric and adult individuals with rare diseases. This survey will be administered next year.

Objective 3.3: Develop a list of resources for individuals with rare diseases on how to self-advocate during interactions with health care providers.

Status: Completed

Summary: Health Care Providers Subcommittee members developed a draft list of resources to guide individuals on self-advocacy (Appendix J). These resources include education, planning for medical appointments, and encouraging testimonials.

Recommended Strategies for State Agencies

Recommendation 1: Identify data needs necessary to understand the population with rare diseases in Florida.

Year 1 Objectives

Objective 1.1: Define incidence of rare disease in Florida.

Status: In Progress

Summary: A barrier identified was that information regarding rare diseases is not collected at the state level because most rare diseases are not included in public health surveillance systems. Data collection resources have been identified and a Data Use Agreement (DUA) is routing through the final stages of approval. Data exchange is planned to proceed in year two for completion.

Objective 1.2: Develop a list of rare diseases with an overlay of distribution across the state, beginning with the top 50 most prevalent rare diseases nationally.

Status: In Progress

Summary: A barrier identified was that information regarding rare diseases is not collected at the state level because most rare diseases are not included in public health surveillance systems. Data collection resources have been identified and a DUA is routing through the final stages of approval.

Objective 1.3: Develop a list of International Classification of Diseases, Tenth Edition codes for conditions covered by the Florida Newborn Screening Program.

Status: In Progress

Summary: From the Genetic and Rare Diseases Information Center (GARD), the Department received a list of the top rare diseases in Florida (Appendix K). A list of conditions covered by the Florida Newborn Screening Program (Appendix L) has also been received. In year two, these lists will be compared to identify the rare disease conditions covered by the Florida Newborn Screening Program.

Recommended Strategies for State Agencies

Recommendation 1: Identify data needs necessary to understand the population with rare diseases in Florida.

Year 1 Objectives

Objective 1.4: Identify the business rules for data extraction and research questions, including legal review.

Status: In Progress

Summary: The Department has developed a DUA to obtain access to the data of interest by the State Agencies Subcommittee that is collected by other agencies. This process will continue in year two.

Objective 1.5: Develop a list of resources with an illustrated network of the varying systems of care that serve individuals with rare diseases such as public services, support organizations, educational support systems, employment services, etc.

Status: In Progress

Summary: This objective, though initiated, has been moved to year two for continued planning and development.

Recommended Strategies for State Agencies

Recommendation 2: Identify areas of improvement for the Department on rare diseases.

Year 1 Objectives

Objective 2.1: Identify Department programs that serve the population with rare diseases.

Status: In Progress

Summary: Rare diseases are associated with specific symptoms. The Department has identified programs that serve individuals with rare diseases regardless of their specific diagnosis. Examples include the Florida Newborn Screening Program, Early Steps, and the Disability and Health Program. Next steps include analysis of these programs and the services they provide.

Objective 2.2: Identify the population with rare diseases the Department serves.

Status: In Progress

Summary: By analyzing the programs and services in objective 2.1, additional information will be collected regarding the populations that receive services. This research will continue in year two.

Objective 2.3: Develop a plan to enhance public-private partnerships.

Status: In Progress

Summary: This objective remains in development. The Department engages in public and private partnerships to advance the work of the RDAC. During the reporting year the Department presented at an annual conference with pharmaceutical technology representatives to share the work of the RDAC to illicit feedback.

Recommended Strategies for State Agencies

Recommendation 2: Identify areas of improvement for the Department on rare diseases.

Year 1 Objectives

Objective 2.4: Develop a plan for interagency data sharing agreements regarding the population with rare diseases.

Status: In Progress

Summary: The Department has started drafting a DUA to obtain access to the data collected within other agencies. This process will continue in year two.

Recommended Strategies for State Agencies

Recommendation 3: Identify areas of improvement for Agency for Health Care Administration on rare diseases.

Year 1 Objectives

Objective 3.1: Identify Florida Medicaid data that can be used to determine prevalence of rare diseases in Florida.

Status: Complete

Summary: This information has been identified by the Agency for Health Care Administration (AHCA) and is currently moving through internal verification processes. Once it has received final approval it will be shared with the Department.

Objective 3.2: Identify Florida Medicaid programs that serve the population with rare diseases.

Status: In Progress

Summary: This information has been identified by AHCA and is currently moving through internal verification processes. Once it has received final approval it will be shared with the Department. The completion of this objective will continue into year two.

Objective 3.3: Identify the population with rare diseases in Florida that Medicaid serves and assess its needs.

Status: In Progress

Summary: This information has been identified by AHCA and is currently moving through internal verification processes. Once it has received final approval it will be shared with the Department. The completion of this objective will continue into year two.

Recommended Strategies for State Agencies

Recommendation 3: Identify areas of improvement for AHCA on rare diseases.

Year 1 Objectives

Objective 3.4: Develop a plan to enhance public-private partnerships.

Status: In Progress

Summary: Collaborative efforts will continue with AHCA to enhance partnerships between public and private sectors. This work will continue in year two.

Recommended Strategies for State Agencies

Recommendation 4: Identify areas of improvement for the Florida Department of Education (FDOE) on rare diseases.

Year 1 Objectives

Objective 4.1: Identify FDOE programs that serve the population with rare diseases and identify any barriers or gaps in services.

Status: Completed

Summary: A list of programs provided by FDOE has been developed (Appendix M). Concluding discussions with other bureaus and divisions within FDOE, there have been no service gaps identified. The RDAC will continue to revisit and maintain this list on an ongoing basis. As any barriers or gaps in service are identified, they too will be included.

Objective 4.2: 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.

Status: Completed

Summary: During the completion of this task, an abundance of supportive resources available to staff, students, and families were identified. It was decided that a list of these resources would better serve those searching for additional information (Appendix M). This list will continue to be maintained and updated. This information is anticipated to be provided on the RDAC website.

Recommended Strategies for State Agencies

Recommendation 5: Identify areas of improvement for the Florida Office of Insurance Regulation (OIR) on rare diseases.

Year 1 Objectives

Objective 5.1: OIR will determine what data on rare diseases currently exist through commercial health insurance plans.

Status: In Progress

Summary: A survey has been developed by the OIR and sent to commercial health insurance plan providers. Survey responses have been received and are being reviewed. This and subsequent objectives, will be moved to year two as the individual responsible for oversight of this recommendation and objective is no longer with the State Agencies Subcommittee.

Objective 5.2: Identify the population with rare diseases OIR serves and assess its needs.

Status: In Progress

Summary: Moved to year two.

Objective 5.3: Identify if current surveys are being utilized to collect feedback from health insurance recipients with rare diseases.

Status: In Progress

Summary: Moved to year two.

Objective 5.4: Identify OIR programs that serve the population with rare diseases.

Status: In Progress

Summary: Moved to year two.

Recommended Strategies for State Agencies

Recommendation 5: Identify areas of improvement for the Florida Office of Insurance Regulation (OIR) on rare diseases.

Year 1 Objectives

Objective 5.5: Develop a plan to enhance public-private partnerships.

Status: In Progress

Summary: Moved to year two.

Objective 5.6: Evaluate requirements for insurance benefits not to be discriminatory against insurance recipients with rare diseases.

Status: In Progress

Summary: Moved to year two.

Recommended Strategies for State Agencies

Recommendation 6: Identify areas of improvement for disaster preparedness and pandemic response for individuals with rare diseases in Florida.

Year 1 Objectives

Objective 6.1: Develop partnership with the Department's Division of Emergency Preparedness and Community Support, State Emergency Operations Center and local emergency shelter administrators on the needs of individuals with rare diseases in Florida.

Status: In Progress

Summary: Disaster preparedness resources exist for individuals and families of those with rare diseases. The next step is for the Department to collaborate with local emergency management teams to improve public messaging.

Objective 6.2: Complete a needs assessment of the population with rare diseases in Florida for disaster preparedness and pandemic response.

Status: In Progress

Summary: Local organizations will be identified to develop a focus group with the purpose of completing a needs assessment of the population with rare diseases. Further development of this objective will be moved into the year two implementation plan.

Conclusion

During the previous year, the RDAC began working toward the completion of the recommendations and objectives they set as goals for year one. The RDAC continues to examine the recommendations and objectives aimed to improve supports and services to individuals with rare diseases in Florida. Through this process, the RDAC identified a need to standardize the processes for the diagnosis of individuals with rare diseases. The RDAC further investigated the incidence and prevalence of rare diseases in Florida while identifying the supportive resources that are currently available for individuals and their families as well as their health care team. Information gathered this past year will provide the foundation for next year progress.

The RDAC remains committed to assisting the rare disease community in Florida and will continue to be a platform and voice for the rare disease community.

Appendix A

Academic Research Institutions Subcommittee Members

A physician who is licensed under Chapter 458 or Chapter 459 and practicing in this state with experience in treating rare diseases:
Barry Byrne, MD, PhD
Director, Powell Gene Therapy Center
Associate Chair, Department of Pediatrics
Professor, Pediatrics and Molecular Genetics & Microbiology
University of Florida

A representative from an academic research institution in Florida, which receives grant funding for research regarding rare diseases:
Divya Patel, DO, MBA
Program Director, Interstitial Lung Disease
Program Director, Sarcoidosis
Associate Professor of Medicine
University of Florida

A representative from an academic research institution in this state which receives grant funding for research regarding rare diseases:
Mustafa Tekin, MD
Division Director, Clinical and Translational Genetics
University of Miami

A physician who is licensed under Chapter 458 or Chapter 459 and practicing in this state with experience in treating rare diseases:
Rajan Wadhawan, MD, MMM
Senior Executive Officer
AdventHealth for Children
AdventHealth for Women

Appendix A

Health Care Provider Subcommittee Members

A representative of organizations in this state which provide care or other support to individuals with rare diseases:
Anita Davis, PT, DPT, FNCP, CNPT
Physical Therapist
Brooks Rehabilitation

A representative of organizations in this state which provide care or other support to individuals with rare diseases:
Rebekah Dorr
Director of Clinical Patient Advocacy
Myasthenia Gravis Hope Foundation

An individual who is, or was previously, a caregiver for individuals with a rare disease:
Zana Dupee, JD
Caregiver

An individual who is 18 years of age or older who has a rare disease:
Jessica O'Reilly, JD
Self-Advocate

An individual who is 18 years of age or older who has a rare disease:
India Steinbaugh, MPH
Self-Advocate

Appendix A

State Agencies Subcommittee Members

An Individual who is a caregiver of an individual with a rare disease:
Eric Biernacki

A representative of the Agency for Health Care Administration:
Ann Dalton, MM
Bureau Chief, Bureau of Medicaid Policy
Florida Agency for Health Care Administration

A representative of the Department of Education:
Kathryn Hebda, MM - RDAC Vice Chair
Chancellor, Florida College System
Florida Department of Education

A representative of the biotechnology industry:
Jonathan Hawayek, MBA
Head of State Government Affairs
Spark Therapeutics, Inc.

A representative of the Department of Health:
Melissa Jordan, MS, MPH - RDAC Chair
Assistant Deputy Secretary for Health
Florida Department of Health

A representative of health insurance companies:
Scott McClelland, PharmD
Vice President, Commercial and
Specialty Pharmacy Programs and
Health Care Solutions
Florida Blue

An individual who is, or was previously, a caregiver for individuals with a rare disease:
Jennifer Sutherland
Caregiver

Appendix B

NORD Rare Disease Centers of Excellence (NORD RD CoE) Program Description

Each of the currently designated NORD RD CoE sites was selected because it has an extensive array of experts covering all medical specialties for both children and adults. The sites have demonstrated a strong commitment to supporting all stages of rare disease research, with the goals of increasing our knowledge of disease etiology and pathophysiology, improving diagnostic techniques, and developing rare disease treatments and cures. In addition to dedication to increasing research and care, each Director/Co-Director and NORD RD CoE site is also dedicated to mentoring the next generation of rare disease clinicians/researchers. The Directors and NORD RD CoE sites view the NORD RD CoE network as a powerful mechanism to engage and train fellows and junior faculty early in their careers.

Appendix C

Framework for Florida Designation of Rare Disease CoE

Criteria	Academic RDRI	Clinical RDRI
Basic Research	Yes*	No
Translational Research	Yes*	No
**Clinical Research (Clinical Trials)	Yes	Yes
Accreditation Council of Graduate Medical Education (ACGME) Programs in Genetics or Accredited Genetic Counseling Training Program	Yes*	No
Rare Disease Designations or Accreditation	Yes	Yes
Rare Disease Education for Clinicians	Yes	Yes
Rare Disease Education for Patients and Families	Yes	Yes
Rare Disease Education for Community	Yes	Yes
Transitional Care from Pediatrics to Adult Care	Yes	Yes
Innovative Rare Disease Programs	Yes	Yes
**Notable Internal/External Rare Disease Collaborations	Yes	Yes
Focus on Patient Satisfaction and Improvement Opportunities	Yes	Yes
**Patient and Family Support Services	Yes	Yes
Performs Health Cost and Benefits Analysis	Yes	Yes

*Only 1 of the 3 starred criteria need to be met (Basic Research, Translational Research, ACGME Genetics or Genetic Counseling Training Program) to be designated as an Academic RDI, while meeting all other criteria.
** Not mandatory.

Scoring Determination of Criteria

Using the criteria in the above table, assign a score of 0, 1, or 2 for each.

- 0 – No
- 1 – Partial
- 2 – Full

These institutions will be working closely with state organizations and work in this area is associated with patients who historically have much higher than average per capita health care costs. By including a criterion of analysis of health costs and benefits, a better understanding of its association with interventions and programs focused on rare disease patient cohorts will be recognized.

Two pivotal questions regarding the above framework will be:

1) Who will evaluate/score the criteria for different institutions?
2) Who will make the final determination for applying institutions?

Appendix D

Florida Pediatric Rheumatologists

Miami

Dr. Arielle D Hay
Practice Name: Chief, Section of Rheumatology, Nicklaus Children's Pediatric Specialists
Primary Office: 3100 SW 62 Avenue #304, Miami, FL 33155
Clinical Interests: Hypermobility Syndrome, juvenile arthritis, juvenile dermatomyositis, juvenile systemic lupus erythematosus and transition of care for adolescents with chronic illness

Dr. Yonit Sterba Rakovchik
Practice Name: Pediatric Rheumatologist, Nicklaus Children's Pediatric Specialists
Primary Office: 3100 SW 62 Avenue #304, Miami, FL 33155
Clinical Interests: Childhood-onset lupus, juvenile idiopathic arthritis (JIA), juvenile dermatomyositis and transition of care

Dr. Rafael F Rivas-Chacon
Practice Name: Director, Division of Rheumatology, Nicklaus Children's Pediatric Specialists
Primary Office: 3200 SW 60 Court #105, Miami, FL 33155
Clinical Interests: Pediatric rheumatology

Dr. Reuven Bromberg
Practice Name: Baptist Health
Primary Office: 715 Southwest 73rd Avenue, Miami, FL 33144
Clinical Interests: Pediatric rheumatology

Boca Raton

Dr. Sandra M. Lawrence
Practice Name: Broward Health Medical Center
Primary Office: 9960 Central Park Blvd North, Suite 225, Boca Raton, FL 33428
Clinical Interests: Children with chronic and life-threating rheumatic diseases such as Kawasaki disease, scleroderma, systematic lupus and more.

Dr. Kristina M. Wiers-Shamir
Practice Name: Joe DiMaggio Children's Hospital.
Primary Office: 900 Glades Road, Boca Raton, FL 33431
Clinical Interests: Pediatric rheumatology, patients with systemic lupus erythematosus, JIA/juvenile rheumatoid arthritis, juvenile dermatomyositis, periodic fever syndromes. Behcet's disease, Henoch-Schonlein purpura, Kawasaki disease, vasculitis, psoriatic arthritis, acute rheumatic fever, post-streptococcal reactive arthritis, hypermobility syndrome, uveitis, chronic non-bacterial osteomyelitis/chronic recurrent osteomyelitis, and other rheumatologic conditions.

Appendix D

Florida Pediatric Rheumatologists

St. Petersburg

Dr. Amanda Schlefman
Practice Name: Johns Hopkins All Children's Hospital
Primary Office: 601 5th St. S, St. Petersburg, FL 33701
Clinical Interests: Pediatric Rheumatology

Dr. Diana S. Milojevic
Practice Name: Johns Hopkins All Children's Hospital
Primary Office: 601 5th St. S, St. Petersburg, FL 33701
Clinical Interests: JIA, systemic lupus erythematosus, juvenile dermatomyositis, scleroderma, vasculitis, periodic fever syndromes and other systemic inflammatory diseases

Dr. Ann M Szymanski
Practice Name: Johns Hopkins All Children's Hospital
Primary Office: 601 5th St. S, St. Petersburg, FL 33701
Clinical Interests: Pediatric Rheumatology, and her research interests have included systemic juvenile idiopathic arthritis

Dr. Laisa M. Santiago-Ramos
Practice Name: Vice Chief of Pediatric Rheumatology
at Johns Hopkins All Children's Hospital
Primary Office: 601 5th St. S, St. Petersburg, FL 33701
Clinical Interests: systemic lupus erythematosus, vasculitis, juvenile dermatomyositis, scleroderma, JIA, Sjogren's syndrome, periodic fever syndromes, and other systemic inflammatory diseases

Orlando

Dr. Kathleen M. Vazzana
Practice Name: Orlando Health Arnold Palmer Hospital for Children
Primary Office: 1802 Kuhl Ave. Ste. 101, Orlando, FL 32806
Clinical Interests: Systemic lupus erythematosus, pediatric rheumatology

Dr. Monica L. Friedman
Practice Name: Orlando Health Arnold Palmer Hospital for Children
Primary Office: 1802 Kuhl Ave. Ste. 101, Orlando, FL 32806
Clinical Interests: JIA, pediatric rheumatology

Appendix D

Florida Pediatric Rheumatologists

Dr. Akaluck Thatayatikom
Practice Name: Advent Health
Primary Office: 2501 N Orange Avenue, Suite 586, Orlando, FL 32804
Clinical Interests: JIA, systemic lupus erythematous, Sjogren's syndrome, dermatomyositis

Dr. Mary Toth
Practice Name: Nemours Children's Health
Primary Office: 6353 Nemours Parkway, Orlando, FL
Clinical Interests: JIA, juvenile rheumatoid arthritis

Pensacola

Dr. Brandon Dorion
Practice Name: Nemours Children's Health, Pensacola
Primary Office: 8331 N. Davis Highway, Pensacola, FL 32514
Clinical Interests: JIA, juvenile rheumatoid arthritis

Lakeland

Dr. Mary Toth (listed above)
Practice Name: Nemours Children's Specialty Care, Lakeland
Primary Office: 1324 Lakeland Hills Blvd Suite A, Lakeland, FL 33803
Clinical Interests: JIA, juvenile rheumatoid arthritis

Gainesville

Dr. Leandra C. Woolnough
Practice Name: UF Health
Primary Office: 1549 Gale Lemerand Drive, Second floor, Gainesville, FL 32610
Clinical Interests: JIA, arthritis, dermatomyositis, scleroderma, systemic lupus erythematosus

Dr. Melissa Elder
Practice Name: UF Health
Primary Office: 1600 SW Archer Road, Gainesville, FL 32610
Clinical Interests: Dermatomyositis, scleroderma, systemic lupus erythematosus, granulomatosis with polyangiitis, rheumatoid arthritis

Appendix D

Florida Pediatric Rheumatologists

Dr. Renee Modica
Primary Office: 1600 SW Archer Road, Gainesville, FL 32610
Practice Name: UF Health
Clinical Interests: JIA, dermatomyositis, scleroderma, systemic lupus erythematosus

Jacksonville

Dr. Jennifer L. Rammel
Practice Name: UF Health Jacksonville
Primary Office: 841 Prudential Dr, Jacksonville, FL 32207
Clinical Interests: Pediatric rheumatology in the critical care setting and management of rheumatology emergencies

Dr. Taha Moussa
Practice Name: UF Health Jacksonville
Primary Office: 841 Prudential Dr, Jacksonville, FL 32207
Clinical Interests: Pediatric rheumatology

Appendix E

Florida Geneticists

Provider Name	Affiliated Institution
Paldeep Atwal	Atwal Clinic
Deborah Barbouth	University of Miami
Ana Maria Rodriguez Barreto	Nicklaus Children's Hospital
Mislen Bauer	Miami Children's Hospital
Paul Benke	Joe DiMaggio Hospital
Ariel Brautbar	Joe DiMaggio Hospital
Amarilis Chancez-Valle	University of South Florida
Ingrid Christian	Orlando Health Arnold Palmer Hospital for Children
Melissa Crenshaw	John Hopkins All Children's Hospital
Majed Dasouki	Advent Health
Irman Forghani	University of Miami
Cheryl Garganta	University of South Florida
Christopher Griffith	University of South Florida
Meral Gunay-Aygun	John Hopkins All Children's Hospital
Jeffrey Innis	Lee Health
Parul Jayakar	Nicklaus Children's Hospital
Sajel Kana	Nicklaus Children's Hospital
Alexander Kim	John Hopkins All Children's Hospital
Kumarie Latchman	University of Miami
Laura Martin	Tallahassee Memorial Hospital
Margarita Moreno	Advent Health
Kathleen Pope	University of South Florida
Lauren Pronman	Nicklaus Children's Hospital
Judith Ranells	University of South Florida
Mustafa Tekin	University of Miami
Willa Thorson	University of Miami
Pamela Trapane	Jacksonville University of Florida
Patricia Wheeler	Arnold Palmer Hospital
Klaas Wierenga	Mayo Clinic Jacksonville
Roman Yusupov	Joe DiMaggio Hospital
Roberto Zori	University of Florida

Appendix F

Rare Disease Organizations with Continuing Medical Education (CME) Resources

Organization	Resource
National Organization for Rare Disorders	https://rarediseases.org/for-clinicians-and-researchers/resources/cme/ Video library https://rarediseases.org/get-involved/educate/educational-initiatives/for-medical-professionals/ Rare disease reports Physician guide for select conditions
National Institutes of Health- Genetic and Rare Disease Information Center	https://rarediseases.info.nih.gov/diseases Browse by disease
Orphanet+A36	https://www.orpha.net/consor/cgi-bin/index.php Inventory of orphan drugs Directory of patient organizations Directory of professionals and institutions Directory of expert centers Directory of laboratories providing diagnostic testing Directory of ongoing research Collection of thematic reports
FindZebra	https://www.findzebra.com/ Search by disease
Eurordis: Europe Rare Disease	https://www.eurordis.org/on-your-disease/ (Links back to Orphanet)
ICORD: International Collaboration on Rare Disease	http://icord.es/ (Links back to Orphanet, GARD and other organizations) (Links back to Food and Drug Administration (FDA) and European Medicines Agency) (Links to patient groups and research)
RSDSA: Reflex Sympathetic Dystrophy Syndrome Association	https://rsds.org/ Clinical guidelines Treatments Educational presentations Course on Complex regional pain syndrome (CRPS) Medical library

Appendix F

Rare Disease Educational Resources for Health Care Providers

Current CME Providers	Website
American College of Physicians	acponline.org
BMJ education and BMJ learning	new-learning.bmj.com
Cleveland Clinic Center for Continuing Education	clevelandclinicmeded.com
Coursera CME courses	coursera.org/courseraplus
Edx CME courses	edx.org
Evidence Based (EB) Medicine	ebmedicine.net
Harvard Medical School	cmecatalog.hms.harvard.edu/
Intiva Health	intivahealth.com
MASTER CLINICIANS	masterclinicians.thinkific.com
Mayo Clinic Online CME	ce.mayo.edu/online-education
Medscape	medscape.org medscape.org/sites/advances/rare-diseases
MER primary care	mer.org
MomMD	mommd.com/cme-categories/
myCME and associates	mycme.com
New England Journal of Medicine (NEJM)	nejm.org/learning?query=main_nav_lg
RELIAS Media	reliasmedia.com
Stanford Center for Continuing Medical Education	med.stanford.edu/cme.html
STATPEARLS	statpearls.com

Appendix G

Patient Centered Care Resources

Articles

Morel, T., Cano, S.J. Measuring what matters to rare disease patients – reflections on the work by the IRDiRC taskforce on patient-centered outcome measures. *Orphanet J Rare Dis* **12**, 171 (2017). https://doi.org/10.1186/s13023-017-0718-x

National Institutes of Health (NIH)

This list of articles is focused on patient-centered care provided by the National Institutes of Health.
https://search.nih.gov/search?utf8=%E2%9C%93&affiliate=nih&query=patient+centered+care&commit=Search

Agency for Healthcare Research and Quality (AHRQ)

Shared Decisionmaking

The Shared decsionmaking model encourages individuals to be assertive when it comes to medical decisions that affect their health.
https://www.ahrq.gov/cahps/quality-improvement/improvement-guide/6-strategies-for-improving/communication/strategy6i-shared-decisionmaking.html

The SHARE Approach - Achieving Patient-Centered Care with Shared Decision making: A Brief for Administrators and Practice Leaders (AHRQ)

The SHARE Approach is a 1-day training program developed by the Agency for Healthcare Research and Quality (AHRQ) to help health care professionals work with patients to make the best possible health care decisions.
https://www.ahrq.gov/health-literacy/professional-training/shared-decision/tool/resource-9.html

Patient Centered Outcomes Research Institute (PCORI)

PCORI presents findings from systematic reviews and some of their funded research studies in concise, accessible formats called Evidence Updates. These updates, which capture the highlights and context for these new findings, are created and disseminated in collaboration with patient, provider, or other organizations. Most Evidence Updates are available in two versions: one for patients and caregivers and one for clinicians and other professionals.

PCORI also provides evidence synthesis reports and interactive visualizations. When patients, clinicians, or other healthcare stakeholders raise a question that does not already have an evidence-based answer, sometimes new research is needed to answer it. But in other cases, a careful look at results from research studies already completed can more quickly provide findings that can be used to make informed choices. PCORI's Evidence Synthesis Initiative takes that approach.

https://www.pcori.org/impact/evidence-synthesis-reports-and-interactive-visualizations

Appendix G

Patient Centered Care Resources

Continuing Education Courses

University of Massachusetts Medical School Center for Integrated Primary Care Certificate Program in Patient-Centered healthcare
This course takes a patient-centered and team-based approach to develop knowledge about patient engagement and care coordination unique to healthcare settings, with an emphasis on supporting patient autonomy and agency within a complex healthcare landscape.
https://www.umassmed.edu/cipc/continuing-education/PatientCenteredHealthcare/

Institute for Healthcare Improvement
Locate learning opportunities provided by the Institute for Healthcare Improvement
https://www.ihi.org/Topics/PFCC/Pages/Education.aspx

Patent and Family Centered Care and Engagement
This seven-minute video is provided by Johns Hopkins School of Medicine. It explains the process of moving from patient and family centered care to engaged and activated care models.
https://www.coursera.org/lecture/patient-safety-culture/patient-and-family-centered-care-and-engagement-0Awxl

Appendix H

Educational Resources on Health Care Practices

National Certification Commission for Acupuncture and Oriental Medicine (NCCAOM)
https://www.nccaom.org/find-a-practitioner-directory/

Environmental Working Group
www.ewg.org
This is an online resource for providers, caregivers and patients that provides information about toxicity levels in specific food, personal care, and home cleaning brands. This site also provides a database for identifying contaminants found in tap water based on ZIP codes.

Johns Hopkins Medicine: Food and Nutrition
https://www.hopkinsmedicine.org/health/wellness-and-prevention/food-and- nutrition
This webpage provides tips for the general public on how to eat healthier while facing challenges such as a limited budget and a fast-paced lifestyle with limited time for food prep.

Appendix I

Medical Practices and Specialists Accepting Pediatric and Adult Patients with Rare Diseases

Jacksonville Health and Transition Services
UF Department of Pediatrics
https://pediatrics.med.jax.ufl.edu/jaxhats/

Powell Center for Rare Disease Research and Therapy
University of Florida
https://raredisease.powellcenter.med.ufl.edu/about-us/#:

Appendix J

Self-Advocacy Resources

Rare Action Network
https://rareaction.org
NORD encourages individuals with rare diseases to connect with other individuals through the Rare Action Network. The Rare Action Network provides access for individuals with rare diseases to research their symptoms before their doctor appointments, gather facts related to their conditions, develop questions to ask their health care providers, and discuss genetic testing, experimental drug trial therapy, and costs for the treatment options. Additionally, the Rare Action Network serves to connect individuals with rare diseases across the country as a support system.

Patient Assistance Programs
https://rarediseases.org/patient-assistance-programs/
The patient assistance programs provided by NORD are designed to help individuals diagnosed with a rare disease to receive financial assistance, caregiver aid, emergency relief and educational support.

Patient Testimonials
https://ncats.nih.gov/funding/challenges/rare-deseases-challenge-2020
The National Institutes of Health (NIH) Division of Rare Diseases Research Innovation offered the “Rare Diseases Are Not Rare!” Challenge to connect individuals with rare diseases nationwide and to share the importance of research and education for treatment methods. Each participant submitted testimonials in the form of art, social media, videos, poems, and personal stories. The Challenge ran from 2018 to 2020 and showcased three winners and five honorable mentions here: https://ncats.nih.gov/funding/challenges/rare-deseases-challenge-2020/winners

NORD also provides patient stories here:
https://rarediseases.org/for-patients-and-families/connect-others/patient-stories/

Appendix K

GARD List of Top Rare Diseases

Rare Disease	GARD ICD-10	Updated ICD-10
Glutaryl-CoA dehydrogenase deficiency	E72.3	
• Short chain acyl CoA dehydrogenase deficiency		E71.312
• Long chain/very long chain acyl CoA dehydrogenase deficiency		E71.310
• Medium chain acyl CoA dehydrogenase deficiency		E71.311
Propionic acidemia	E71.1	E71.121
Alpha-1-antitrypsin deficiency	E88.0	E88.01
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	G11.1	G11.8
Behçet disease	M35.2	M35.2
Brugada syndrome	I49.8	
• Unspecified right bundle-branch block		I45.1
• Other specified cardiac arrhythmias		I49.8
• Other specified congenital malformations of heart		Q24.8
Systemic primary carnitine deficiency	E71.3; E71.41	E71.41
Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy	G60.0	G60.0
Cartilage-hair hypoplasia	Q78.8	Q78.8
Sickle cell anemia	D57.0; D57.1; D57.2	
• Sickle cell disease without crisis		D57.1
• Sickle-cell thalassemia, unspecified, with crisis		D57.419
• Sickle-cell thalassemia without crisis		D57.40
• Sickle-cell/Hb-C disease without crisis		D57.20
• Other sickle-cell disorders without crisis		D57.80
• Sickle-cell/Hb-C disease with crisis, unspecified		D57.219
• Sickle-cell/Hb-C disease with splenic sequestration		D57.212
• Sickle-cell thalassemia, unspecified, with acute chest syndrome		D57.411
• Sickle-cell thalassemia, unspecified, with splenic sequestration		D57.412
• Sickle-cell thalassemia beta zero with crisis, unspecified		D57.439
• Other sickle-cell disorders with crisis, unspecified		D57.819
• Sickle-cell thalassemia beta zero with crisis		D57.43
• Sickle-cell thalassemia beta plus with crisis		D57.45
• Other sickle-cell disorders with acute chest syndrome		D57.811
• Sickle-cell thalassemia beta plus with crisis, unspecified		D57.459
• Sickle-cell thalassemia, unspecified, with cerebral vascular involvement		D57.413
• Sickle-cell thalassemia beta zero without crisis		D57.42
• Sickle-cell thalassemia beta zero with splenic sequestration		D57.432
• Sickle-cell thalassemia beta plus without crisis		D57.44
• Sickle-cell thalassemia beta plus with splenic sequestration		D57.452

Appendix K

GARD List of Top Rare Diseases (Infectious Diseases Removed)

Rare Disease	GARD ICD-10	Updated ICD-10
• Other sickle-cell disorders with cerebral vascular involvement		D57.813
• Other sickle-cell disorders with crisis with other specified complication		D57.818
• Sickle-cell/Hb-C disease with crisis with other specified complication		D57.218
• Sickle-cell thalassemia, unspecified, with crisis with other specified complication		D57.418
• Sickle-cell thalassemia beta zero with acute chest syndrome		D57.431
• Sickle-cell thalassemia beta plus with acute chest syndrome		D57.451
• Sickle-cell thalassemia beta plus with crisis with other specified complication		D57.458
• Other sickle-cell disorders with crisis		D57.81
Oculopharyngeal muscular dystrophy	G71.0	G7.09
Steinert myotonic dystrophy	G71.1	G71.11
Ellis Van Creveld syndrome	Q77.6	Q77.6
Multiple osteochondromas	Q78.6	Q78.6
Familial Mediterranean fever	E85.0	M04.1
Congenital hypothyroidism	E00.0, E00.1, E00.2, E03.1, E00.9	
• Congenital hypothyroidism without goiter		E03.1
• Congenital hypothyroidism with diffuse goiter		E03.0
Rare cutaneous lupus erythematosus	L93.0, L93.1, L93.2	
• Other local lupus erythematosus		L93.2
• Subacute cutaneous lupus erythematosus		L93.1
• Discoid lupus erythematous		L93.0
MELAS	G71.3	E88.41
Peripartum cardiomyopathy	O90.3	O90.3
GNE myopathy	G71.8	G71.2
Distal myopathy, Welander type	G71.0	G71.2
Noonan syndrome	Q87.1	Q87.19
46, XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	E29.1	Q99.0
Alpha-thalassemia	D56.0	D56.0
Fetal and neonatal alloimmune thrombocytopenia	P61.0	P61.0
Turner syndrome	Q96.0, Q96.1, Q96.8, Q96.9, Q96.2, Q96.3, Q96.4, 96.0	
• Karotype 46, x iso		Q96.1

Appendix K

GARD List of Top Rare Diseases (Infectious Diseases Removed)

Rare Disease	GARD ICD-10	Updated ICD-10
• Other variants of Turner syndrome		Q96.8
• Turner's syndrome, unspecified		Q96.9
• Karotype 46, x abnormal sex characteristics except iso (Xq)		Q96.2
• Mosaicism, 45, X/46, XX or XY		Q96.3
• Mosaicism, 45, X/other cell line(s) with abnormal sex chromosome		Q96.4
Hydrops fetalis	P83.2, P56.0, P56.9	
• Hydrops fetalis NOS		P83.2
• Hydrops fetalis due to isoimmunization		P56.0
• Hydrops fetalis due to other and unspecified hemolytic disease		P56.9
• Hydrops fetalis due to unspecified hemolytic disease		P56.90
• Hydrops fetalis due to other hemolytic disease		P56.99
Isolated anencephaly/exencephaly	Q00.0	Q00.0
Bowen-Conradi syndrome	Q87.8	Q87.89
Aorta coarctation	Q25.1	Q25.1
Dermatitis herpetiformis	L13.0	
• Duhring-Brocq disease		L13.0
• Juvenile dermatitis herpetiformis		L12.2
• Senile dermatitis herpetiformis		L12.0
Hemoglobin C disease	D58.2	D58.2
Holoprosencephaly	Q04.2	Q04.2
Tetralogy of Fallot	Q21.3	
• Tetralogy of Fallot		Q21.3
• Personal history of other corrected congenital malformations		Z87.79
• Personal history of (corrected) congenital malformations of heart and circulatory system		Z87.74
Vernal keratoconjunctivitis	H16.2	
• Vernal keratoconjunctivitis		H16.26
• Vernal keratoconjunctivitis, with limbar and corneal involvement, right eye		H16.261
• Vernal keratoconjunctivitis, with limbar and corneal involvement, left eye		H16.262
• Vernal keratoconjunctivitis, with limbar and corneal involvement, bilateral		H16.263
• Vernal keratoconjunctivitis, with limbar and corneal involvement, unspecified eye		H16.269

Appendix K

GARD List of Top Rare Diseases (Infectious Diseases Removed)

Rare Disease	GARD ICD-10	Updated ICD-10
Cholangiocarcinoma	C24.8, C24.9, C22.1, C24.0	
• Intrahepatic bile duct carcinoma		C22.1
• Personal history of malignant neoplasm of other digestive organs		Z85.09
• Malignant neoplasm of overlapping sites of biliary tract		C24.8
• Malignant neoplasm of biliary tract, unspecified		C24.9
• Malignant neoplasm of extrahepatic bile duct		C24.0
Oculocutaneous albinism type 2	E70.3	
• Oculocutaneous albinism		E70.32
• Chediak-Higashi syndrome		E70.330
• Hermansky-Pudlak syndrome		E70.331
• Oculocutaneous albinism, unspecified		E70.329
• Tyrosinase negative oculocutaneous albinism		E70.320
• Tyrosinase positive oculocutaneous albinism		E70.321
• Other oculocutaneous albinism		E70.328
• Amelogenesis imperfecta	K00.5	K00.5
Systemic sclerosis	M34.0, M34.1, M34.2, M34.8, M34.9	
• Systemic sclerosis , unspecified		M34.9
• Systemic sclerosis with polyneuropathy		M34.83
• Systemic sclerosis with myopathy		M34.82
• Systemic sclerosis with lung involvement		M34.81
• Systemic sclerosis induced by drug and chemical		M34.2
• CR(E)ST syndrome		M34.1
• Progressive systemic sclerosis		M34.0
Autosomal dominant optic atrophy	H47.2	
• Unspecified optic atrophy		H47.20
• Hereditary optic atrophy		H47.22
• Primary optic atrophy, bilateral		H47.213
• Other optic atrophy, bilateral		H47.293
• Primary optic atrophy, right eye		H47.211
• Primary optic atrophy, left eye		H47.212

Appendix K

GARD List of Top Rare Diseases (Infectious Diseases Removed)

Rare Disease	GARD ICD-10	Updated ICD-10
• Primary optic atrophy, unspecified eye		H47.219
Amish nemaline myopathy	G71.2	G71.21
Amish lethal microcephaly	Q02	Q02
Charcot-Marie-Tooth disease type 1A	G60.0	G60.0
Trehalase deficiency	E74.3	E74.39
Myotonic dystrophy	G71.1	G71.11

Note: Original sources for rare diseases were Genetic and Rare Diseases Information Center (GARD). ICD-10 codes were refined using the rare diseases as search terms on ICD-10 data.com and including the supplied, relevant ICD-10 codes. In the table, the original codes are in blue and the updated codes with ICD-10 mapping are in green. In addition, the bolded terms are from the original rare disease list supplied by GARD, and the indented terms underneath the bolded terms are additional codes that were found to be related to the bolded term after further ICD-10 mapping. Please note that this list has not been validated or reviewed by rare disease experts. It may produce false positives.

Appendix L

Newborn Screening ICD-10 Codes

Core Conditions	ICD-10 Codes
Organic Acid	
• Propionic Acidemia (PROP)	E71.121
• Methylmalonic Acidemia (Methylmalonyl-CoA Mutase Deficiency) (MUT)	E71.120
• Methylmalonic Acidemia (Cobalamin Conditions)	E71.120
• Isovaleric Acidemia (IVA)	E71.110
• 3-Methylcrotonyl-CaA Carboxylase Deficiency (3-MCC)	E71.19
• 3-Hydroxy-3-Methylglutaric Aciduria (HMG)	E71.118
• Holocarboxylase Synthetase deficiency (MCD)	D81.818
• Beta-Ketothiolase Deficiency (BKT)	D81.818
• Glutaric Acidemia, Type I (GA-1)	E72.3
Other	
• Biotinidase Deficiency (BIOT)	D81.810
• Critical Congenital Heart Disease (CCHD)	Q20.0, Q20.3, Q21.3, Q22.0, Q22.4, Q23.4, Q26.2
• Cystic Fibrosis (CF)	E84
• Classic Galactosemia (GALT)	E74.21
• Hearing Loss or Varying Hearing Levels	H91.90
• Sever Combined Immunodeficiency (SCID)	D81.9
• X-Linked Adrenoleukodystrophy (X-ALD)	E71.520, E71.522, E71.528, E71.529
• Pompe	E74.02
• Spinal Muscular Atrophy (SMA)	G12.9
• Mucopolysaccharidosis, Type I (MPS I)	E76.01
Fatty Acid Oxidation	
• Carnitine Uptake Defect (CUD)	E71.41
• Medium-Chain acyl-CoA Dehydrogenase Deficiency (MCAD)	E71.311
• Very Long-ChainAcyl-CoA Dehydrogenase Deficiency (VLCAD)	E71.310
• Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)	E71.318
• Trifunctional Protein Deficiency (TFP)	E71.318
Amino Acid	
• Argininosuccinic Aciduria (ASA)	E72.22
• Citrullinemia, Type I (CIT-I)	E72.23
• Maple Syrup Urine Disease (MSUD)	E71.0
• Homocystinuria (HCY)	E72.11
• Classic Phenylketonuria (PKU)	E70.0
• Tyrosinemia, Type I, (TYR I)	E70.21
Endocrine	
• Primary Congenital Hypothyroidism (CH)	E03.1
• Congenital Adrenal Hyperplasia (CAH)	E25.0
Hemoglobin	
• Sickle Cell Disease (Hemoglobin SS Disease)	D57.1

Appendix L

Newborn Screening ICD-10 Codes

Secondary	ICD-10 Codes
Amino Acid	
• Citrullinemia, Type II (CIT II)	E72.23
• Hypermethioninemia (MET)	E72.19
• Benign Hyperphenylalaninemia (H-PHE)	E70.1
• Biopterin Defect in Cofactor Biosynthesis (BIOPT-BS)	Unknown
• Biopterin Defect in Cofactor Regeneration (BIOPT-REG)	Unknown
• Ornithine Transcarbamylase Deficiency (OTC)	E72.4
• Carbamoyl Phosphate Synthetase Deficiency (CPS)	E72.29
• Tyrosinemia, Type II (TYR II)	E70.21
• Tyrosinemia, Type III (TYR III)	E70.21
Fatty Acid Oxidation	
• Short-Chain Acyl-CoA Dehydrogenase (SCAD)	E71.312
• Glutaric Acidemia, Type II (GA-2)	E71.313
• Carnitine Palmitoyltransferase, Type I (CPT-1A)	E71.318
• Carnitine Palmitoyltransferase, Type II (CPT-II)	E71.318
• Carnitine Acylcarnitine Translocase Deficiency (CACT)	E71.318
Organic Acid	
• Methylmalonic Acidemia with Homocystinuria (Cbl C, D, F)	E71.120
• Isobutyrylglycinuria (IBG)	E71.19
• 2-Methylbutyrylglycinuria (2MGB)	E71.19
• 3-Methylglutaconic Aciduria (3MGA)	E71.111
• 2-Methyl-3Hydroxybutyric (2M3HBA)	E71.19
• Ethylmalonic Encephalopathy (EME)	E88.49
Hemoglobin	
• Hemoglobin C, D, and E Conditions	D58.2
• Various Hemoglobinopathies	Unknown
Other	
• T-cell related Lymphocyte Deficiencies	Unknown

Appendix M

Florida Department of Education Programs and Resources

Audience	Title	Link	Description
Parent/Guardians, Teachers	A Parent & Teacher Guide to Section 504: Frequently asked questins (FAQs)	https://www.fldoe.org/core/fileparse.php/7690/urlt/0070055-504bro.pdf	A short guide to Section 504 with the most frequently asked questions.
Parent/Guardians, Teachers	Office of Healthy Schools	https://www.fldoe.org/schools/healthy-schools/	Webpage to the Florida Department of Education's Office of Healthy Schools, including resources utilized to provide students with the knowledge and skills necessary to be healthy throughout their lifetime.
Parents/ Guardians, Students, Teachers	Individuals with Disabilities Education Act (IDEA)	https://www.fldoe.org/academics/exceptional-student-edu/beess-resources/individuals-with-disabilities-edu-act-/index.stml	Links to the U.S. Department of Education's and the Federal Register's IDEA information.
Parents/ Guardians, Students, Teachers	Educational resources to support exceptional students	https://www.fldoe.org/academics/exceptional-student-edu/	This includes specific educational program information; Parent Information; related services; student employment transition services; eligibility for a variety of supports based on conditions; DOE, school district, and related agency staff contacts
Parents/Guardians	Getting Ready for Your Student's Individualized Education Plan (IEP) Meeting	https://www.fldoe.org/core/fileparse.php/7690/urlt/IEPmeeting.pdf	Document to assist parents/guardians before and during their child's IEP meeting.
Parents/Guardians	Florida School Choice Main Page	https://www.fldoe.org/schools/school-choice/	Includes links and information about all scholarship programs, private schools, charter schools, virtual education and other specific educational choice options

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Florida Department of Education Programs and Resources

Audience	Title	Link	Description
Parents/Guardians	Parent Information for Specific School Choice Options	https://www.fldoe.org/schools/school-choice/parent-resources/	Provides more detailed information and brochures on the most requested choice option information.
Parents/Guardians	Family Empowerment Scholarship for Unique Abilities (K-12)	https://www.fldoe.org/schools/school-choice/k-12-scholarship-programs/fes/index.stml	Webpage describing Florida's K-12 Scholarship that can be used for private school tuition or as an education savings plan. Includes links for applying.
Parents/Guardians, Students	Service Animal Guidelines	http://info.fldoe.org/docushare/dsweb/Get/Document-7346/dps-2015-60a.pdf	Guidelines for the Use of Service Animals by Students with Disabilities.
Parents/Guardians, Students	Division of Blind Services	https://dbs.fldoe.org/	Webpage to the Florida Department of Education's Division of Blind Services webpage which includes resources for parents/guardians and students.
Students	Florida's Workforce Education Initiative Special Populations College Services Resource Guide	https://dbs.fldoe.org/Resources/get-there.html	Allows persons with disabilities to view/access the resources each Florida College System institution provides.
Teachers	Selecting Accommodations: Guidance for IEP Teams	https://www.fldoe.org/core/fileparse.php/7690/url/0070064-selectingaccommodations.pdf	A brief explanation of the process the IEP team should follow to determine which accommodations a student with a disability may need.

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Florida Department of Education Programs and Resources

Audience	Title	Link	Description
Teachers	Designing Lessons for the Diverse Classroom – A Handbook for Teachers	https://www.fldoe.org/core/fileparse.php/7690/url/0070084-4dclessn.pdf	Designed to assist school districts, and the state agencies which support education programs, in the provision of special programs.
Teachers	Curriculum Planning and Learning Management Systems (CPALMS)	https://www.cpalms.org/Public/search/Standard	CPALMS organizes courses and resources based on Florida state adopted standards, including access points. Access points were developed for students with significant disability to access general education content.