Rare Disease Advisory Council

2024 Annual Legislative Report Prepared in Response to Louisiana Revised Statute 40:1122.1

Prepared by:

Bureau of Family Health

Office of Public Health

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LOUISIANA
DEPARTMENT OF HEALTH

Submitted to:

Jeff Landry, Governor, State of Louisiana

Health and Welfare Committee, Louisiana Senate

Health and Welfare Committee, Louisiana House of Representatives

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Acknowledgements:

This report was compiled and written by the Louisiana Department of Health, Office of Public Health, Bureau of Family Health staff responsible for supporting the Rare Disease Advisory Council. Amy Zapata, MPH, is the director of the Bureau of Family Health. Key Bureau of Family Health contributors include Children's Special Healthcare Services Program Manager, Cheryl Harris, and Genetic Diseases Strategy Manager, Rodney Goldsmith.

We recognize the members of the Louisiana Rare Disease Advisory Council and the team at the Bureau of Family Health whose dedication and hard work make this report possible, including the communications staff who edited and designed the report.

Lastly, we honor the people impacted by rare diseases represented in this report. It is our sincere hope that the activities of the Louisiana Rare Disease Advisory Council will make progress in addressing the needs of the rare disease community and work toward a more informed and supportive environment for all those impacted by rare diseases in Louisiana.

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Introduction

The Louisiana Department of Health, Office of Public Health, Bureau of Family Health is responsible for improving the health and well-being of Louisiana residents diagnosed with a rare disease by promoting research and sharing vital information to ensure equitable access to treatments and resources needed to live a fulfilling life. Overall, the Bureau works to promote the health of Louisiana families throughout their lifetime by implementing programs and initiatives aimed at improving the health of pregnant women, babies, children, teens, adults, and youth with special healthcare needs.

Our vision is for Louisiana to be a state where all people are empowered to reach their full potential, from birth through the next generation. Our mission is to elevate the strengths and voices of individuals, families, and communities to catalyze transformational change to improve population health and achieve equity. The Rare Disease Advisory Council and this report serve as tools for the Bureau of Family Health to advance maternal and child health outcomes within the state.

Rare Diseases

A rare disease is a disease or condition that affects fewer than 200,000 people in the U.S.[1] Over 6,800 types of rare diseases exist, affecting an estimated 25 million to 30 million people.[2] This translates to approximately 10% of the total national population.[3] While particular diseases may be rare, the total number of people living with rare diseases is significant. For several decades, the rare disease community has used this estimate to illustrate the significant impact of rare diseases.

Rare diseases are a significant contributor to chronic illness, disability, and premature death in children and adults. They represent a major cause of long-term health issues and can lead to considerable hardship and loss of life across all age groups. The complexity and often costly, ineffective treatments of rare diseases place a disproportionate burden on healthcare resources and create significant administrative demands.

The number of individuals living with a rare disease in Louisiana remains unknown due to the lack of an established methodology for identifying the incidence and prevalence of these diseases. In addition, the lack of quality data makes it challenging to identify new cases, geographic regions of increased risk, the most affected groups, or hospital utilization.

Rare Disease Advisory Council

The National Organization for Rare Diseases defines a rare disease advisory council (RDAC) as a platform that empowers the rare disease community to advocate more effectively within state government. RDACs address the needs of patients with rare diseases and their families by enabling external partners to recommend solutions to state leaders on critical issues experienced by these individuals. These include the need for increased awareness, diagnostic tools, and access to affordable treatments and cures.[4]

In 2015, the first state advisory council on rare diseases was created in North Carolina. Since the inception of the first RDAC, several states have established their own advisory councils. The national trend is taking hold, with rare disease

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[1] (Public 97-414 97th Congress an Act)

[2] (Rare Diseases FAQ, 2024)

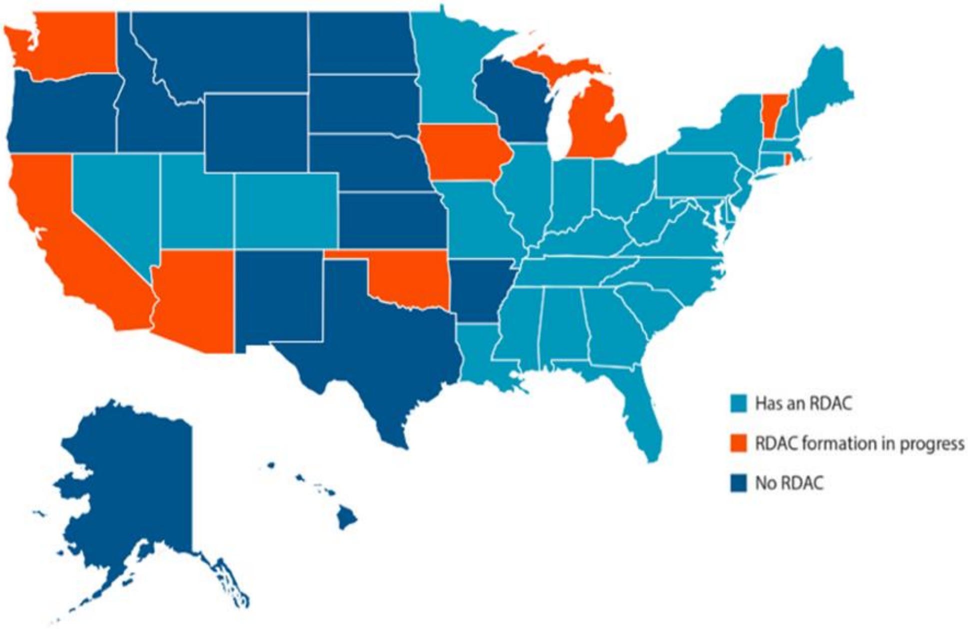
[3] (About Genetic and Rare Diseases (GARD) Information Center, 2024)

[4] (Rare Disease Advisory Councils, 2024)

stakeholders across the country diligently working to establish a mechanism that allows improved representation of this historically underserved and often misunderstood population.

Enacted in 2021, Louisiana Revised Statute (R.S.) 40:1122.1 (see Appendix C) established the Louisiana Rare Disease Advisory Council, making Louisiana the 18th state with a rare disease council. Nationally, 28 established RDACs (see Figure 1) support the rare disease community and demonstrate that RDACs can be an invaluable resource for individuals diagnosed with a rare disease and their families.[5]

Figure 1: Rare Disease Advisory Councils by State.[6]



As a statutorily mandated advisory group, the Louisiana Rare Disease Advisory Council provides resources to public and private agencies in the state that serve individuals diagnosed with rare diseases. The Council is tasked with compiling an annual report that summarizes the findings, activities, and progress for State Fiscal Year 2024 (SFY 2024) of the Council and recommendations for addressing the needs of Louisianans living with a rare disease. The Council operates under the Bureau of Family Health, which provides staff support and policy expertise to assist the Council in fulfilling its mission to provide advice on research, diagnosis, treatment, and education related to rare diseases. The Council's additional duties, outlined in R.S. 40:1122.1 (see Appendix D), include providing expert and clinical advice to the Drug Utilization Review Board in its review of treatments for rare diseases and providing input and feedback to the Louisiana Department of Health and other agencies on matters affecting people with rare diseases.

Findings, Activities and Progress

While the Louisiana Rare Disease Advisory Council is still in its early stages of development, members (listed in Appendix A) are dedicated to improving treatment, care, and support for individuals diagnosed with a rare disease in Louisiana. In SFY 2024, the Council held three general meetings (see more in Appendix B). Additionally, the Council chair met with Bureau of Family Health staff on a bimonthly basis to review legislative requirements, create meeting

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[5] (Rare Disease Advisory Councils, 2024)

[6] (RDACs by State, 2024)

agendas, and address the Council's administrative needs. These include record-keeping, scheduling, conducting research, and tracking the progress of tasks or projects assigned at meetings.

The Louisiana Rare Disease Advisory Council reviewed an implementation toolkit and webinars from the National Organization for Rare Disorders and its Project RDAC to inform the development of its operational guidelines and a work plan to carry out the Council's charges. The following information provides status updates on the activities required by law, as established by R.S. 40:1122.1 (see Appendix C for the full statute).

Recommendations

The Louisiana Rare Disease Advisory Council is legislatively mandated to research the patient population in the state, assess access to coverage, care and unmet needs, promote awareness and education, and identify and address emergency preparedness and response needs. In SFY 2024, the Council consulted with physicians and leaders from other state RDACs to identify the needs of individuals with rare diseases and develop recommendations for improving service provider awareness and education. The Council's following recommendations will help improve the landscape for Louisiana's rare disease population through collaborative efforts, informed advocacy, and the provision of valuable resources.

1. **Collaborations with Local and National Advocacy Groups**
    a. Develop and promote awareness campaigns, webinars, data collection surveys, and workshops to facilitate knowledge-sharing opportunities between experts in rare diseases and healthcare providers. The Louisiana Rare Diseases Advisory Council recommends that the Louisiana Department of Health create materials to promote Rare Disease Day on February 28 of each year.
    b. Use resources offered by the National Organization for Rare Diseases and support the Council with developing and disseminating user-friendly toolkits, treatment guides, and case studies focusing on rare diseases. The Louisiana Rare Diseases Advisory Council recommends that the Louisiana Department of Health promote Rare Disease Day based on the national toolkit messaging.

2. **Advocacy for Policy Changes**
   The Louisiana Rare Diseases Advisory Council recommends that the Louisiana Department of Health, policymakers, and relevant organizations engage in policy reform to ensure that resources and education regarding rare diseases are prioritized and adequately funded. The Council requests the Louisiana Department of Health to review and update current resources and education for publication on the Rare Diseases Advisory Council website on an annual basis.

3. **Peer Education**
   The Louisiana Rare Diseases Advisory Council recommends that the Louisiana Department of Health identify platforms and forums where healthcare providers specializing in rare diseases can share their expertise with colleagues. The Council requests the Louisiana Department of Health to assist the Council with producing educational materials and content for an annual webinar on a rare disease topic of interest.

   In addition, the Louisiana Rare Diseases Advisory Council recommends that the Louisiana Department of Health identify platforms and forums where individuals and families affected by rare diseases can connect with one another, share personal stories and experiences, and offer support to others.

Priorities

Understanding the Rare Disease Populations in Louisiana and Their Needs

Data is essential to understanding who is affected by rare diseases and illuminating the actions needed to support their care and quality of life. The number of individuals living with a rare disease in Louisiana remains unknown due to the lack of established methodology for identifying the incidence and prevalence of rare diseases.[*] In SFY 2024, the Louisiana Rare Disease Advisory Council met with rare disease advocates, physicians, and leaders from RDACs around the country to help identify effective methods to determine the population and needs.

Establishing a system for estimating the number of people living with rare diseases in the state is one of the Council's most significant priorities. In SFY 2025, the Council will collaborate with the Bureau of Family Health's staff to identify sound methods to quantify and monitor rare diseases and the number of people affected using existing data sources. Anticipated data sources available through public health include health information (i.e. ICD-10 and ICD-11 disease codes) from Medicaid, Medicare, major hospital systems, and managed care organizations. The aim is to establish a reliable and replicable method for estimating the population and evaluating access to specialty care, therapies, supports, insurance, and disability benefits for those affected by rare disorders. Bureau of Family Health staff will collaborate with the Office of Public Health's Bureau of Health Informatics and Louisiana Medicaid to conduct preliminary analyses of hospital discharge and Medicaid claims data. Council members will review the data and provide feedback and recommendations to state agencies and the Louisiana Department of Health to ensure patient needs are properly addressed.

In SFY 2024, the Louisiana Rare Disease Advisory Council proposed a survey for SFY 2025 to collect data from rare disease patients and their healthcare providers, aiming to better quantify and understand their needs. The community members' surveys will help identify the complexity and magnitude of challenges associated with currently available resources, programs, and support. The providers' surveys will help determine the support needed to deliver high-quality medical services to individuals with rare diseases. These assessments will be distributed through rare disease support organizations, major hospital systems, and social media. Bureau of Family Health staff will assist with survey development, distribution, and analysis of its findings.

Education and Resources

For Providers: The Louisiana Rare Disease Advisory Council consulted with rare diseases advisory councils in other states about developing awareness campaigns and educational initiatives, and discussed various ways to support the medical community regarding resources. Topics included implementing a compendium of organizations that provide resources to individuals with rare diseases and establishing the Extension for Community Healthcare Outcomes (ECHO) model to inform community practitioners in virtual settings on how to treat patients with rare conditions.

For State Leaders and Policymakers: One of the Council's key functions is to provide comprehensive education on rare disease research, education, diagnosis, and treatment to elected officials, state leaders, and government officials. With the aim of serving as a unified voice for the rare disease community, in SFY 2025, the Council plans to engage these constituencies to share challenges experienced by individuals with rare conditions, including challenges related to legislation and policies affecting people with rare diseases. The Council will also assess membership composition to ensure all appropriate perspectives are represented. Bureau of Family Health staff will assist the Council in conducting outreach to constituencies whose perspective informs the work and efforts of the Council.

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[*] Incidence is defined by the number of new cases that develop in a specified time, and the proportion of a population with a specific characteristic in a specified timeframe determines prevalence.

For Individuals and Families Affected by Rare Disease: The Louisiana Rare Disease Advisory Council requested the creation of a website and a social media account for the purpose of sharing council-related information and announcements. In response, the Bureau of Family Health leveraged its established social media channels and website to provide content and helpful resources to increase the public's knowledge of rare diseases and the impacts on a person living with a rare disease.

Provide Expert and Clinical Advice to the Drug Utilization Board in its Review of Treatments

The authorizing statute for the Louisiana Rare Disease Advisory Council charges the Council with working with the Louisiana Department of Health Drug Utilization Board to provide expertise and clinical advice. Work related to this charge was not initiated in SFY 2024, but will be pursued in SFY 2025.

Conclusion

The Louisiana Rare Disease Advisory Council was established to serve as a resource to public and private agencies in Louisiana that provide services to individuals diagnosed with rare diseases. In SFY 2024, the Council made significant progress in its early stages; however, there is still much work to do to ensure that individuals diagnosed with a rare disease in the state receive the care they need. For SFY 2025, the Council will continue focusing on implementing the strategies previously mentioned to advance service provider awareness and education related to rare diseases. Looking ahead, the Council is poised to continue making significant progress in addressing the needs of the rare disease community and working toward a more informed and supportive environment for all those impacted by rare diseases in Louisiana.

Appendix A: Louisiana Rare Disease Advisory Council Membership Roster

Member's Name	Role	Appointed By
Libby Airhart, Chair *(resigned as chair on 12/07/23)*	A caregiver for a person diagnosed with a rare disease	Governor
Kelly Rouse, Vice-Chair *(appointed chair on 12/07/23)*	Louisiana House of Representatives	Speaker of the House of Representatives
Meredith Allain, MD *(resigned on 10/27/23)*	A geneticist who is licensed and practicing in this state	Governor
Hans Andersson, MD	A physician who is licensed and practicing in this state with experience in researching, diagnosing, or treating rare diseases	Governor
Marissa Benchea	A resident of this state who is 18 years of age or older and has been diagnosed with a rare disease	Governor
Jennifer Borrillo	A representative from an academic research institution in this state that conducts rare disease research	Governor
Deiadra Garrett, MD	A physician who is licensed and practicing in this state with experience in researching, diagnosing, or treating rare diseases	Governor
Jonathan James	Louisiana Senate	President of the Senate
Ross Klingsberg, MD	A representative from an academic research institution in this state that conducts rare disease research	Governor
Carolyn Moore, FNP-BC	A registered nurse or advanced practice registered nurse who is licensed and practicing in this state and has experience treating rare diseases	Governor
Lauren Williams	A representative of a rare disease patient organization operating in this state	Governor
Vacant	A representative from the Department	Governor

Appendix B: Louisiana Rare Disease Advisory Council Meeting Schedule

The SFY 2024 Louisiana Rare Disease Advisory Council meeting schedule is listed below, with links to each meeting's agenda and minutes. The agendas and minutes for prior meetings are located on the Bureau of Family Health Boards and Commissions webpage at ldh.la.gov/page/Boards-Commissions-Councils-Family-Health-Louisiana in the Rare Disease Advisory Council section.

- October 24, 2023: Agenda | Minutes
- February 6, 2024: Agenda | Minutes
- May 7, 2024: Agenda | Minutes

Appendix C: Revised Statute 40:1122.1

§1122.1. Louisiana Rare Disease Advisory Council[7]

A. For the purpose of this Part, the following terms have the meanings ascribed to them.

1) “Board” means the Drug Utilization Review board established by the Louisiana Department of Health.

2) “Department” means the Louisiana Department of Health.

3) “Rare disease” means any disease or condition that affects fewer than two hundred thousand persons in the United States. Rare disease shall also mean sickle cell disease and sarcoidosis.

B. 1) The Louisiana Rare Disease Advisory Council is hereby created within the Department of Health:

2) The Louisiana Rare Disease Advisory Council, referred to hereafter in this Part as the “advisory council,” shall serve only in a resource capacity to any public and private agency located in this state that provides services for a person who has been diagnosed with a rare disease.

3) The advisory council shall be composed of the following members:
   a. One member appointed by the president of the Senate.
   b. One member appointed by the speaker of the House of Representatives.
   c. Members appointed by the governor as follows:
      i. One representative from the department.
      ii. Two representatives from academic research institutions in this state that conduct rare disease research.
      iii. Two physicians who are licensed and practicing in this state with experience in researching, diagnosing, or treating rare diseases.
      iv. One geneticist who is licensed and practicing in this state.
      v. One registered nurse or advanced practice registered nurse who is licensed and practicing in this state and has experience treating rare diseases.
      vi. Two residents of this state who are eighteen years of age or older and who have either been diagnosed with a rare disease or is a caregiver for a person who has been diagnosed with a rare disease.
      vii. One representative of a rare disease patient organization operating in this state.

4) The governor shall determine who serves as the chair and vice chair of the advisory council.

5) To the extent practicable, every organization or entity that provides a nomination to the advisory council shall strive for diversity in its appointment on the basis of race, ethnicity, sex, professional or educational background, and geographic residency.

C. The advisory council shall hold its initial meeting no later than October 1, 2021. The advisory council shall meet at least quarterly in a calendar year and at any other time as it deems necessary.

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[7] (Louisiana Revised Statute 40:1122.1. Acts 2021, No. 321, §2; Acts 2022, No. 271, §4., 2024)

D. The purposes of the advisory council include all of the following:

1) To provide input and feedback to the department and any other state agency on matters that affect a person who has been diagnosed with a rare disease, including but not limited to all of the following:
    (a) Pandemic preparedness and response and its impact on a person living with a rare disease.
    (b) Research, education, diagnosis, and treatment relating to rare diseases within this state.
    (c) Rare diseases in general, the severity of rare diseases, and unmet medical needs associated with rare diseases.
    (d) The demographics and clinical description of patient populations.
    (e) Timely access to screening, care, insurance or Medicaid coverage, specialists, and other needed services for a patient who has been diagnosed with a rare disease.
    (f) The impact that coverage, cost-sharing, tiering and any other utilization management procedure has on providing treatment and services to a patient who has been diagnosed with a rare disease.
2) To provide expert and clinical advice to the board in its review of treatments for a rare disease. The treatments may include drugs or biological products emerging from fields of personalized medicine and non-inheritable gene editing therapeutics
3) To provide a report to the governor, the Legislature of Louisiana, the department, and any other relevant agency of both of the following:
    (a) Any findings, activities, and progress of the advisory council pursuant to the provisions of Paragraphs (1) and (2) of this Subsection.
    (b) Any recommendations for addressing the needs of a person living with a rare disease in this state.

E. The advisory council shall not have authority on any matter relating to the department or the board.

F. Nothing in this Section shall require the board to follow the recommendations of the advisory council.

G. Nothing in this Section shall require the advisory council, the board, or any state agency to consult with a person on any matter or be required to meet with any specific expert or stakeholder.

H. An advisory council member shall not receive any compensation for serving on the advisory council.

References

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