

# Rare Disease Advisory Council

*State Fiscal Year 2025*

*Annual Legislative Report*

*In Accordance with R.S. 40:1122.1*

*Prepared by:*

**Bureau of Family Health**

*Office of Public Health*

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**Submitted to:**

- Jeff Landry, Governor, State of Louisiana
- Patrick McMath, Chair, Health and Welfare Committee, Louisiana Senate
- Dustin Miller, Chair, Health and Welfare Committee, Louisiana House of Representatives
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**Acknowledgements:**

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We recognize the members of the Louisiana Rare Disease Advisory Council, whose dedication and hard work made this report possible, and the other Bureau of Family Health staff, who contributed to the report.

Lastly, we honor the people living with a rare disease 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.

## Table of Contents

Executive Summary.....	4
Introduction .....	5
Rare Diseases .....	5
Rare Disease Advisory Council .....	6
Membership.....	6
Pandemic/Emergency Preparedness and Impact on Persons with a Rare Disease.....	7
Research, Education, Diagnosis, and Treatment of Rare Diseases .....	8
Overview of Rare Diseases and Unmet Needs.....	9
Patient Demographics, Clinical Data, Access to Care and Services, and Cost Impact on Treatment .....	10
Expert Guidance on Treatment Reviews.....	10
Conclusion.....	11
Appendix .....	12
Appendix A: Council Membership .....	12
Appendix B: Summary of Council Meetings.....	13
Appendix C: Revised Statute 40:1122.1 .....	14
Appendix D: Rare Disease Data.....	16
Appendix E: Louisiana’s State Report Card .....	17
Bibliography .....	18

## Executive Summary

The Louisiana Rare Disease Advisory Council (LRDAC) serves as a resource to any public and private agency located in Louisiana that provides services for an individual diagnosed with a rare disease.<sup>1</sup> The Council is tasked with compiling an annual report, as outlined in R.S. 40:1122.1, that summarizes the findings, activities, and recommendations for addressing the needs of residents living with a rare disease. This report reflects the findings, activities, and recommendations of the LRDAC for state fiscal year (SFY) 2025.

The Council operates under the Bureau of Family Health (BFH), which provides staff support and policy expertise to assist the Council in fulfilling its mission to provide expert and clinical advice to the [Drug Utilization Review Board](#)<sup>2</sup> on treatments for rare diseases. Additionally, the Council offers input and feedback to the Louisiana Department of Health (LDH) and any other state agencies on issues affecting individuals diagnosed with a rare disease, including but not limited to the following:

- Pandemic preparedness and response, and its impact on a person living with a rare disease;
- Research, education, diagnosis, and treatment, relating to rare diseases;
- Rare diseases in general, the severity of rare diseases, and unmet medical needs associated with rare diseases;
- The demographics and clinical description of patient populations;
- 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; and
- The impact that coverage, cost-sharing, tiering, and any other utilization management procedures have on providing treatment and services to a patient who has been diagnosed with a rare disease.

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<sup>1</sup> (Louisiana Revised Statute 40:1122.1. Acts 2021, No. 321, §2; Acts 2022, No. 271, §4., 2024, 2025).

<sup>2</sup> (Rare Disease Advisory Councils, 2025)

## Introduction

The Louisiana Department of Health (LDH), Office of Public Health (OPH), Bureau of Family Health (BFH), is responsible for improving the health and well-being of residents living 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. The bureau's broader mission is to promote the health of families throughout their lifetime through programs and initiatives to support 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 valued 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. The Genetic Diseases Program, the Rare Disease Advisory Council, and this report are some of the ways BFH works to advance maternal and child health outcomes in the state.

## Rare Diseases

The Food and Drug Administration defines a rare disease as a condition affecting fewer than 200,000 people in the United States.<sup>3</sup> Over 10,000 types of rare diseases exist, affecting an estimated 25 million to 30 million people.<sup>4 5</sup> While particular diseases may be rare, the total number of people living with a diagnosed rare disease is significant. For several decades, the rare disease community has used this estimate to illustrate the significant impact of rare diseases. Examples of rare diseases are cystic fibrosis, which affects the respiratory and digestive systems; Huntington's disease, which affects the brain and nervous system; and muscular dystrophies, which affect the muscles.<sup>6</sup>

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 nature of treatments for rare diseases, which may be ineffective, place a disproportionate burden on healthcare resources and create significant administrative demands.<sup>7</sup>

The number of individuals living with a rare disease 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.

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<sup>3</sup> (Orphan Drug Act [House Resolution 5238], 97th Congress, 1983).

<sup>4</sup> (FAQs about Rare Diseases, 2025).

<sup>5</sup> (Rare Diseases FAQ, 2025).

<sup>6</sup> (FAQs about Rare Diseases, 2025).

<sup>7</sup> (Tisdale, 2021).



## Activities, Progress, and Recommendations

In accordance to Louisiana R.S. 40:1122.1 (D)(1), the LRDAC is charged with giving input and feedback to LDH and other state agencies on matters that affect individuals living with a rare disease. This report describes the activities and progress of the Council during SFY 2025. It also provides recommendations to help improve the landscape for Louisiana's rare disease population through collaborative efforts, informed advocacy, and the provision of valuable resources.

### Pandemic/Emergency Preparedness and Impact on Persons with a Rare Disease

As a marginalized group of the population, individuals diagnosed with a rare disease do not receive the attention they deserve, even outside of a pandemic or emergency situation such as a natural disaster.<sup>10</sup> During a pandemic or emergency situation, they may experience significant delays in receiving a diagnosis, may receive multiple misdiagnoses, and have limited effective treatment options. The Louisiana Rare Disease Advisory Council presents the following recommendations to ensure stakeholders are adequately prepared to assist individuals diagnosed with a rare disease during a pandemic or emergency.

#### **Recommendation to create a template of an emergency medical information document for individuals living with a rare disease**

The Louisiana Rare Disease Advisory Council recommends that LDH create a comprehensive emergency medical information template to assist individuals diagnosed with a rare disease. This template should include essential details such as personal information, caregiver and medical team contacts, current medications, allergies, medical history, and specific emergency treatment instructions. The purpose of this document is to provide first responders and healthcare providers with critical information during emergencies, ensuring that individuals receive appropriate and timely care, even when personnel are unfamiliar with their rare condition. Being that this document contains personal health information, the individual with a rare disease or caregiver is responsible for providing pertinent information and sharing the document with first responders and healthcare providers.

The recommendation is that a checklist is created to assist patients in the event they are incapacitated. The patient and/or caregiver has the responsibility of determining how first responders are made aware of the checklist.

#### **Recommendation to develop and share a comprehensive contact list of emergency medical facilities for individuals living with rare diseases**

The Louisiana Rare Disease Advisory Council recommends that the LDH and/or agencies providing services for individuals living with a rare disease create a comprehensive contact list of local medical facilities and emergency agencies. This list should be available to individuals living with a rare disease and their families to support them when implementing emergency preparedness procedures and accessing resources within their region. The resource list should include:

- A list of emergency facilities capable of treating individuals living with rare diseases;
- Established procedures for notifying local and regional first responders about individuals with critical medical needs during power outages, natural disasters, or other emergencies; and
- Contact information for relevant agencies responsible for critical care and support services.

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<sup>10</sup> (Chowdhury, 2021).

This information should be on the LDH website and other relevant platforms to ensure accessibility. The goal is to enhance coordination, awareness, and emergency response capabilities, ensuring individuals living with rare diseases receive appropriate and timely care during emergencies.

### **Recommendation to ensure pandemic/emergency preparedness plans developed by schools address students with specific healthcare needs**

It is recommended that all public and private schools develop and implement comprehensive pandemic preparedness plans tailored to the healthcare needs of individual students. These plans should include strategies to address each child's specific requirements during emergencies, such as safe evacuation procedures and the provision of individualized support throughout the duration of the emergency while on school premises. Ensuring that these customized plans are in place will help protect vulnerable students and promote a safer, more responsive school environment during pandemics and other health emergencies.

## **Research, Education, Diagnosis, and Treatment of Rare Diseases**

### **The Inaugural Rare Disease Day at the Capitol**

On February 28, 2025, the Louisiana Rare Disease Advisory Council, in partnership with the Louisiana Arts and Science Museum, Baton Rouge Metropolitan Airport, Baton Rouge Zoo, and more than a dozen providers, patient organizations, and vendors, hosted the first annual Rare Disease Day at the Capitol. The event took place in the Louisiana State Capitol Formal Gardens in Baton Rouge and aimed to promote awareness, community engagement, and connection within the rare disease community across the state.

Council members actively promoted the event through social media platforms coverage in local and national news outlets, including [WAFB news](#),<sup>11</sup> [Louisiana First News](#),<sup>12</sup> and [Red River Radio](#).<sup>13</sup> [Lieutenant Governor Billy Nungesser](#) also endorsed the event.<sup>14</sup> Louisiana Rare Disease Advisory Council chair Kelly Rouse served as the master of ceremonies and introduced the following guest speakers: Jonathan James, president and CEO of Hope Charities; Justina Plowden, Miss Wheelchair Louisiana; Nikhia Sims, Miss Louisiana State University; and Eva Delatte, Miss Southeastern University. Over 300 participants from around the state attended the event, including individuals with rare diseases, advocates, state government officials, industry representatives, and medical providers. The Council has already begun planning for the 2026 Rare Disease Day at the Capitol.

### **Public and Administrative Meetings**

In SFY 2025, the Louisiana Rare Disease Advisory Council held four general meetings and eight planning meetings. Throughout the year, the Council discussed strategies to address significant delays in appointments and improve communication with individuals recruited for Council membership regarding the status of their applications. Other key topics included reviewing the Council's by-laws, establishing a webpage and a social media account to share resources and announcements related to rare diseases, and aligning [RS 40:1121.1](#) with the practices of other rare disease advisory councils across the country.

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<sup>11</sup> (James, 2025)

<sup>12</sup> (Rouse, Rare Disease Day at Louisiana State Capitol has educational resources, activities, 2025)

<sup>13</sup> (Rouse, Looking out for the Zebras: advocating for the Rare Disease Community in Louisiana, 2025)

<sup>14</sup> (Nungesser, 2025)

The Louisiana Rare Disease Advisory Council also served as a public platform for members of the rare disease community and advocates to share their lived experiences. Many shared challenges, such as traveling long distances for treatment, visiting multiple specialists due to initial misdiagnoses, and receiving guidance on treatment options and resources for caregivers and patients alike.

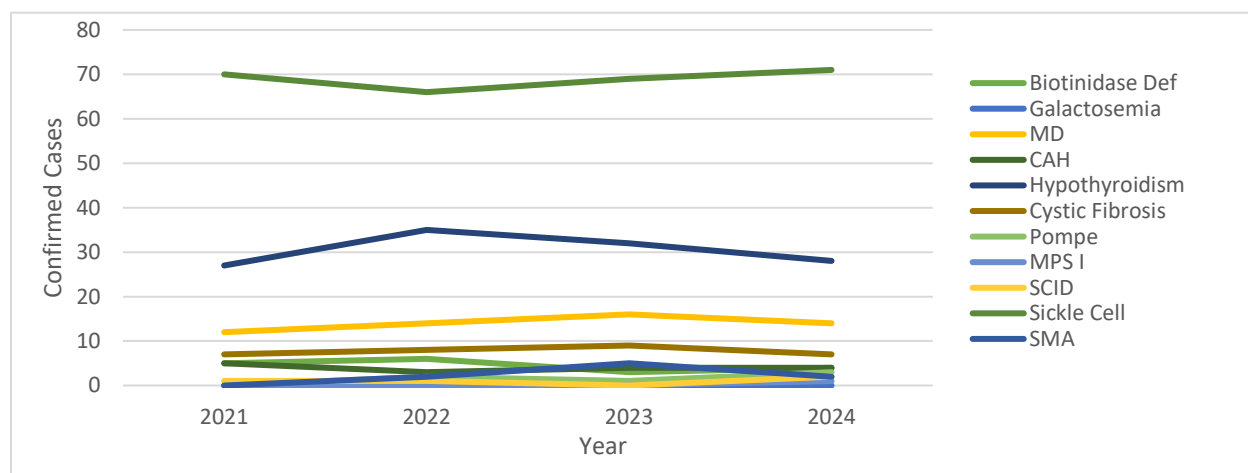
### Overview of Rare Diseases and Unmet Needs

Rare diseases frequently fall beyond the range of mainstream medical research and development, resulting in gaps in diagnosis, treatments, and systems of care coordination. Many patients often have to endure a lengthy, discouraging process waiting for an accurate diagnosis.<sup>15</sup> Even after receiving a diagnosis, patients and their families may have limited or no access to appropriate treatments, leaving them with few options for effective care and management.<sup>16</sup>

Most rare diseases pose serious health risks, often threatening lives and leading to major illness. Despite advances in scientific understanding of their causes and progression, fewer than 5% of rare diseases have approved treatments.<sup>17</sup>

The majority of rare diseases affect children.<sup>18</sup> Newborn screening plays a vital role in identifying certain rare diseases early and enabling prompt treatment that can help prevent disabilities and ensure children receive the support they need at an early age.<sup>19</sup> [Figure 2](#) illustrates the number of confirmed rare disease cases detected through the [Newborn Screening Program](#) between 2021 and 2025 (additional data is provided in [Appendix D](#)).

**Figure 2: Confirmed Rare Disease Cases through Newborn Screening (2021-2024)<sup>20</sup>**



In 2024, BFH’s Newborn Screening Program received a rating of “B” by the National Organization for Rare Disorders.<sup>21</sup> The National Organization for Rare Disorders State Report Card evaluates each state using a

<sup>15</sup> (Phillips, 2024)

<sup>16</sup> (Bauskis, 2025)

<sup>17</sup> (FAQs about Rare Diseases, 2025).

<sup>18</sup> (Rare Disease Challenges in Each Step of the Drug Development Process - Video, 2025).

<sup>19</sup> (Louisiana Newborn Screening Program, 2025).

<sup>20</sup> (Newborn Screening Program, 2025).

<sup>21</sup> (Louisiana State Report Card, 2025).

letter grade system (A-F) based on whether they have adopted policies that are key to operating a successful and impactful newborn screening program. Louisiana’s “B” rating reflects continued progress toward strengthening early detection and intervention for infants with rare diseases.

Since 2021, BFH has screened over 500 newborns with potentially rare conditions. Of those screened, 141 were confirmed to have rare diseases such as spinal muscular atrophy (SMA), severe combined immunodeficiency (SCID), Pompe disease, and hypothyroidism. Identifying these rare diseases early through newborn screening allows for earlier intervention and effective care planning.

### **Patient Demographics, Clinical Data, Access to Care and Services, and Cost Impact on Treatment**

In SFY 2025, LRDAC members attended the National Organization for Rare Disorders’ Rare Disease Breakthrough Summit in Washington, D.C., engaging in sessions with thought leaders from patient advocacy organizations, industry, academia, research, and government. These discussions focused on the timeliest and critical issues facing the rare disease community.

LRDAC plans to collaborate with BFH staff to develop effective methods for measuring and monitoring rare diseases and the affected populations using existing data sources. Some of the existing data sources include ICD-10 and ICD-11 disease codes tracked by Medicaid, Medicare, major hospital systems, and managed care organizations. BFH will collaborate with the OPH’s Bureau of Health Informatics and Louisiana Medicaid to conduct preliminary analyses of hospital discharge and Medicaid claims data. The Council members will review the findings, provide feedback, and share recommendations to state agencies and the LDH to ensure patient needs are accurately identified and addressed.

Additionally, LRDAC will redistribute a previously used survey to collect data from individuals living with a rare disease and their healthcare providers to better understand their needs. The data collected will help identify the complexity and scope of challenges related to existing resources, programs, and support systems. Surveys of healthcare providers will help determine the support necessary 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. BFH staff will assist with survey development, distribution, and analysis of the results. In SFY 2026, the Council will continue to develop and refine its recommendations based on these results.

### **Expert Guidance on Treatment Reviews**

The authorizing statute for the Louisiana Rare Disease Advisory Council charges the Council with working with LDH’s [Drug Utilization Review Board](#) to provide expertise and clinical guidance. Although work related to this charge was not initiated in SFY 2025, the Council plans to pursue it in SFY 2026.

## Conclusion

The Louisiana Rare Disease Advisory Council was established to serve as a resource to public and private agencies that provide services to individuals diagnosed with rare diseases. In SFY 2025, the Council made significant progress and plans to continue doing the work needed to ensure that individuals living with a rare disease in the state receive the care they need. For SFY 2026, the Council will continue focusing on implementing the strategies outlined in this report to advance service provider awareness and education related to rare diseases. Looking ahead, the Council is committed to further progress in addressing the needs of the rare disease community and working toward a more informed, supportive environment for all those living with rare diseases.

## Appendix A: Council Membership

Member's Name	Role	Appointed By
<b>Hans Andersson, MD</b>	A physician who is licensed and practicing in this state with experience in researching, diagnosing, or treating rare diseases	Governor
<b>Jennifer Borrillo</b>	A representative from an academic research institution in this state that conducts rare disease research	Governor
<b>Jonathan James</b>	Louisiana Senate	President of the Senate
<b>Ross Klingsberg, MD</b>	A representative from an academic research institution in this state that conducts rare disease research	Governor
<b>Kelly Rouse, Chair</b>	Louisiana House of Representatives	Speaker of the House of Representatives
<b>Lauren Williams</b>	A representative of a rare disease patient organization operating in this state	Governor
<b>Vacant</b>	A geneticist who is licensed and practicing in this state	Governor
<b>Vacant</b>	A resident of this state who is 18 years of age or older and has been diagnosed with a rare disease	Governor
<b>Vacant</b>	A caregiver for a person diagnosed with a rare disease	Governor
<b>Vacant</b>	A physician who is licensed and practicing in this state with experience in researching, diagnosing, or treating rare diseases	Governor
<b>Vacant</b>	A registered nurse or adv. practice registered nurse who is licensed and practicing in this state and has experience treating rare diseases	Governor
<b>Vacant</b>	A representative from the Louisiana Department of Health	Governor

## Appendix B: Summary of Council Meetings

Meeting Type	Date	Meeting Summary
Administrative Meeting	July 19, 2024	
Regular Meeting	August 6, 2024	<a href="#">Agenda</a>   <a href="#">Minutes</a>
Administrative Meeting	August 20, 2024	
Administrative Meeting	November 14, 2024	
Administrative Meeting	November 21, 2024	
Regular Meeting	December 3, 2024	<a href="#">Agenda</a>   <a href="#">Minutes</a>
Administrative Meeting	February 5, 2025	
Regular Meeting	February 12, 2025	<a href="#">Agenda</a>   <a href="#">Minutes</a>
Administrative Meeting	April 8, 2025	
Regular Meeting	April 25, 2025	<a href="#">Agenda</a>   <a href="#">Minutes</a>
Administrative Meeting	May 7, 2025	
Administrative Meeting	June 11, 2025	

## Appendix C: Revised Statute 40:1122.1

### §1122.1. Louisiana Rare Disease Advisory Council<sup>22</sup>

- I. For the purpose of this Part, the following terms have the meanings ascribed to them.
  - A. “Board” means the Drug Utilization Review board established by the Louisiana Department of Health.
  - B. “Department” means the Louisiana Department of Health.
  - C. “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.
- II.
  - A. The Louisiana Rare Disease Advisory Council is hereby created within the Department of Health:
  - B. 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.
  - C. The advisory council shall be composed of the following members:
    1. One member appointed by the president of the Senate.
    2. One member appointed by the speaker of the House of Representatives.
    3. Members appointed by the governor as follows:
      - a. One representative from the department.
      - b. Two representatives from academic research institutions in this state that conduct rare disease research.
      - c. Two physicians who are licensed and practicing in this state with experience in researching, diagnosing, or treating rare diseases.
      - d. One geneticist who is licensed and practicing in this state.
      - e. One registered nurse or advanced practice registered nurse who is licensed and practicing in this state and has experience treating rare diseases.
      - f. 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.
      - g. 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.

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<sup>22</sup> (Louisiana Revised Statute 40:1122.1. Acts 2021, No. 321, §2; Acts 2022, No. 271, §4., 2024, 2025).

- III. 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.
- IV. The purposes of the advisory council include all of the following:
  - A. 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:
    - 1. Pandemic preparedness and response and its impact on a person living with a rare disease.
    - 2. Research, education, diagnosis, and treatment relating to rare diseases within this state.
    - 3. Rare diseases in general, the severity of rare diseases, and unmet medical needs associated with rare diseases.
    - 4. The demographics and clinical description of patient populations.
    - 5. 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.
    - 6. 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.
  - B. 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
  - C. To provide a report to the governor, the Legislature of Louisiana, the department, and any other relevant agency of both of the following:
    - 1. Any findings, activities, and progress of the advisory council pursuant to the provisions of Paragraphs (1) and (2) of this Subsection.
    - 2. Any recommendations for addressing the needs of a person living with a rare disease in this state.
- V. The advisory council shall not have authority on any matter relating to the department or the board.
- VI. Nothing in this Section shall require the board to follow the recommendations of the advisory council.
- VII. 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.
- VIII. An advisory council member shall not receive any compensation for serving on the advisory council.

## Appendix D: Rare Disease Data

### Data Identified by the Newborn Screening Program (2021-2024)

The BFH Louisiana Newborn Screening Program screens over 99% of live births and plays a critical role in the early detection genetic, metabolic, or congenital disorders that are present at the time of birth. If a disorder is found and treated early, it is often possible to prevent physical or intellectual delays or early death.<sup>23</sup>

The chart below displays the number of confirmed cases for a selection of these conditions (2021 – 2024), underscoring the value of newborn screening in the rare disease landscape.<sup>24</sup>

Disorder	2021	2022	2023	2024
Biotinidase Deficiency	5	6	3	4
Classic Galactosemia	0	0	0	0
Metabolic Disorder	12	14	16	14
Congenital Adrenal Hyperplasia (CAH)	5	3	4	4
Congenital Hypothyroidism	27	35	32	28
Cystic Fibrosis	7	8	9	7
Pompe Disease	N/A	2	1	3
Mucopolysaccharidosis Type I (MPS I)	N/A	0	0	1
Severe Combined Immunodeficiency (SCID)	1	1	0	2
Sickle Cell Disease (SCD)	70	66	69	71
Spinal Muscular Atrophy (SMA)	N/A	2	5	2

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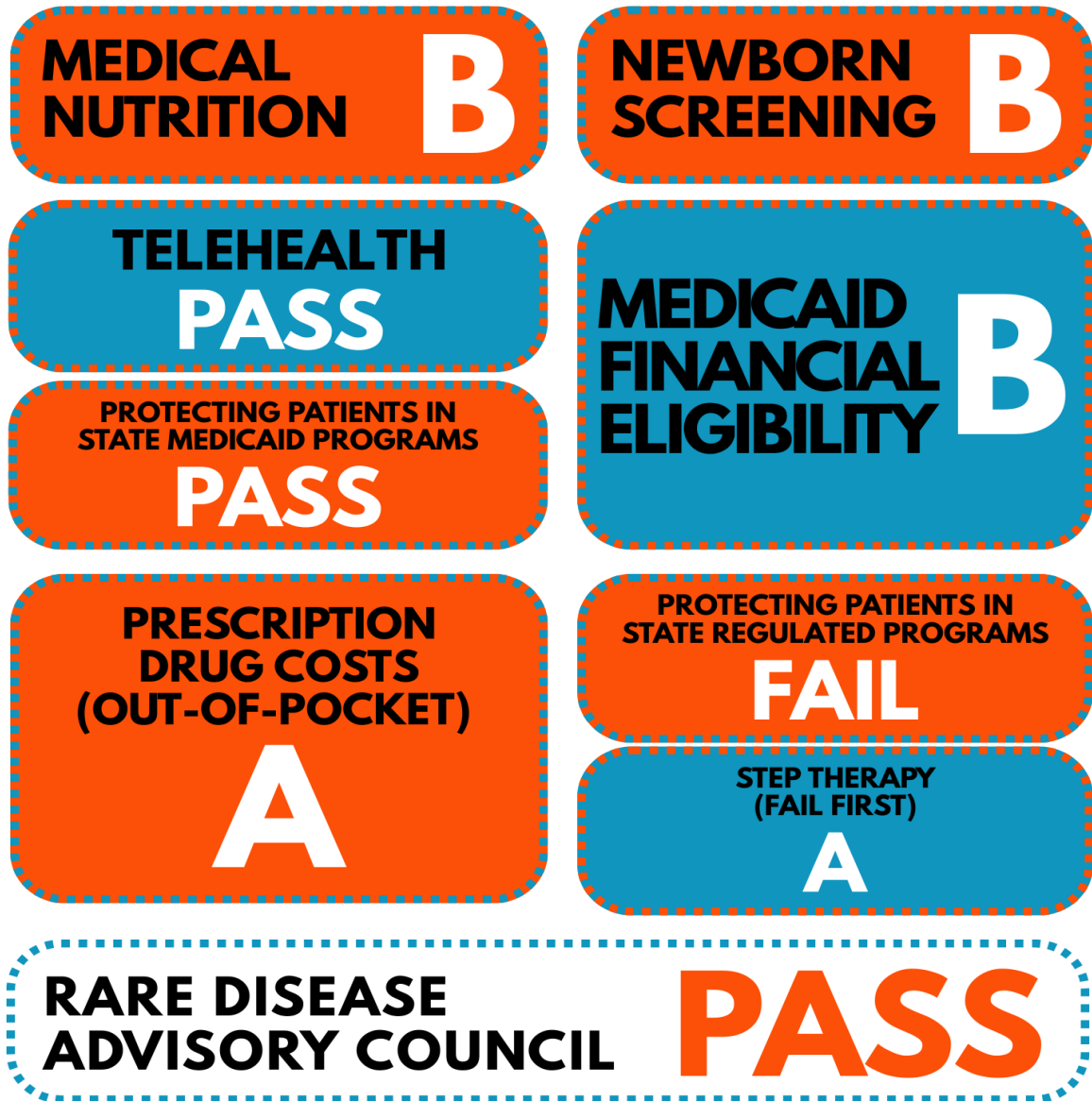
<sup>23</sup> (Louisiana Newborn Screening Program, 2025)

<sup>24</sup> (Newborn Screening Program, 2025).



### LOUISIANA'S CURRENT REPORT CARD

The National Organization for Rare Disorders (NORD) publishes an annual State Report Card that evaluates how well each U.S. state supports people with rare diseases. The report covers several key areas, including newborn screening, access to medical nutrition, and the presence of Rare Disease Advisory Councils (RDACs). The latest edition of the State Report Card was compiled using data current as of November 2024.



<sup>25</sup> (Louisiana State Report Card, 2025).

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