RARE DISEASES...More Common Than You Think

NVRDAC

Rare Disease Advisory Council

STRATEGIC PLAN 2023-2025

www.nvrdac.org

EMPOWERING

Nevadans Living with a

Rare Disease to

Improve Their Future...

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Vision For The Future

Our primary objective is to ensure that the voices of the rare disease patient and the providers who care for them have their voices heard. My name is Annette Logan-Parker, I am the current chair of the Nevada Rare Disease Advisory Council (NV-RDAC) and the Founder & CEO of Cure 4 The Kids Foundation. I am honored to hold the position and am committed to advancing the objectives of the council and fulfilling the duties assigned to us through SB-315 .

The Nevada Rare Disease Advisory Council is pleased to share our 2023-2025 Strategic Plan. We believe that through intentional planning and focused efforts our advisory council can provide a positive impact for the people of Nevada living with a rare disease, as well as for those healthcare professionals working hard to care for them. The NV-RDAC plans to give rare disease patients and providers a much-needed voice in Nevada and our goal is to make it easier for them to articulate their needs and concerns as they sit at the table with state officials.

For years Nevada, as well as many other states, has failed to adequately address rare disease issues. Primarily due to a basic lack of awareness of the patients' and the providers' challenges. The NV-RDAC plans to bridge the gap between patients, providers, and state lawmakers. Rare diseases should be common knowledge.

Nevada's government officials and lawmakers have several powerful policy levers at their disposal. Their ability to create health policies, regulate insurance implementation, and oversee public health gives them the power to transform the lives of rare disease patients. By using these various means strategically and in concert; in complete collaboration with our state government officials, the NV-RDAC plans to advance a more holistic approach toward improving the health and well-being of the rare disease community. The NV-RDAC holds a significant promise for solving some of the many problems faced by thousands of Nevadans, and we will not rest until rare diseases get the attention they deserve.

This strategic plan is designed to serve as the initial framework for creating a collaborative approach to the establishment of a comprehensive plan for the management of rare disease in Nevada. Collaboration and support from numerous government, academic, public health, community-based and other private sector entities will be essential to its success.

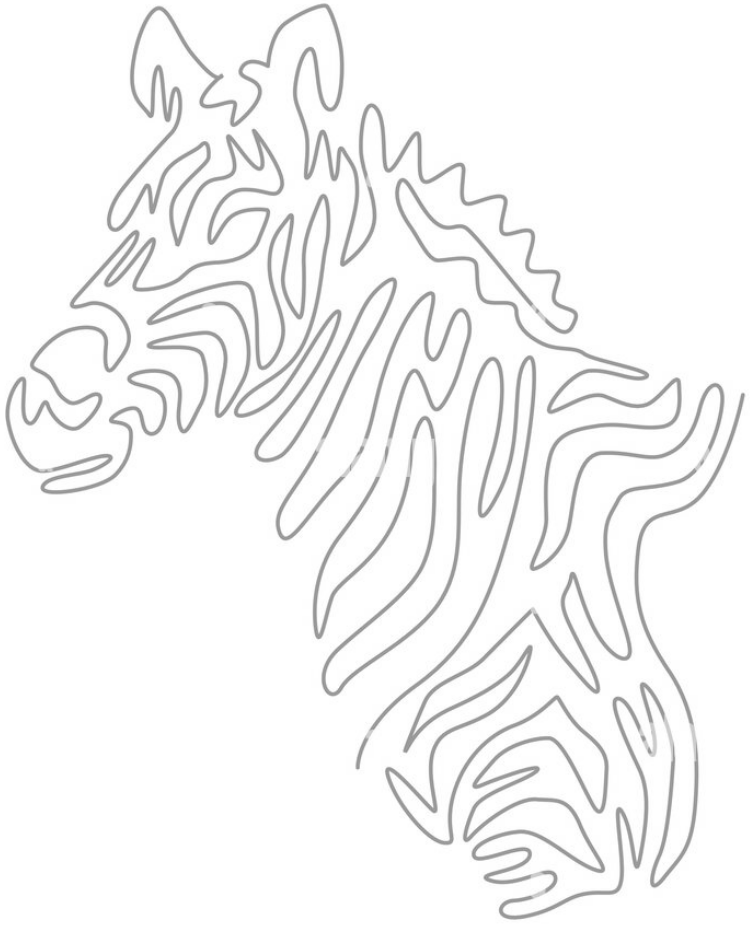
Together we can make rare diseases a little less rare.

Annette Logan-Parker

To Learn More: Please join us for our regularly scheduled meetings.
Meeting schedule: We meet 6 times a year, every other month (February, April, June, August, October, and December) on the first Friday of the month at 9:30 AM (*Time subject to change, please confirm using the QR code below)

Rare Disease Advisory Council Meeting Schedule (nv.gov)

For a list of current RDAC members, please see www.NVRDAC.org.



MISSION

The Nevada Rare Disease Advisory Council's mission is to improve the quality of life and support care for Nevadans affected by rare diseases through collaboration, education, support, and advocacy. Our council seeks to advocate for Nevadans impacted by rare diseases as well as identify the overall impact these diseases have in our community.

What is a RARE DISEASE?

In the late 1940s, Dr. Theodore Woodward, professor at the University of Maryland School of Medicine, instructed his medical interns: "When you hear hoofbeats, think of horses, not zebras." *Rare Disease Report*™ wants to encourage physicians and patients to always keep an eye out for the zebras, too, though. The rare disease community is always growing, but these uncommon conditions can sometimes be difficult to identify. It's always important to embrace your stripes.

These **5 FACTS** are a good place to start.

FACT 1

<200,000

In the United States, a disease or disorder is defined as "rare" when it affects fewer than 200,000 Americans at any given time.

FACT 2

Common symptoms can mask underlying rare diseases and lead to misdiagnosis and treatment delay.

FACT 3

50%

of rare diseases affect children.

FACT 4

The lack of scientific knowledge and quality information on a rare disease often delays a correct diagnosis and can result in heavy burdens, both social and financial, on patients and their families.

FACT 5

80%

of rare diseases have identified genetic origins, while others result from infections (bacterial or viral), allergies, and environmental causes or are degenerative and proliferative.

SOURCES: www.rarediseaseday.org/article/what-is-a-rare-disease

RARE DISEASES ARE MORE COMMON THAT YOU THINK!

1 in 10

Americans has a rare disease + **30 MILLION PEOPLE**

Globally, **50% of those living with a rare disease are CHILDREN**

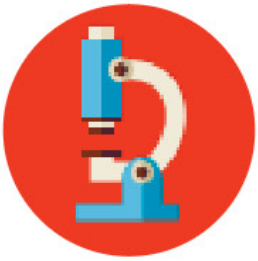
If all people affected by a rare disease lived in one country, this would make up the world's third-most populous nation - equivalent to the total US population

This means more than 300 million people around the world have a rare diseases - approximately the same number of monthyly users of Instagram

80%

of Rare Diseases are Genetically Based

Many Rare Diseases result in premature death of infants and young children or are FATAL in early adulthood



Families & private foundations provide about 3% of ALL medical research funding for Rare Diseases in the US

Introduction to Rare Diseases

In the United States, the Food and Drug Administration (FDA) defines a rare disease as any disease that affects fewer than 200,000 Americans. Our European colleagues consider a disease rare when it affects less than 1 in 2,000 people. Regardless of the contrastingly different attitudes towards what constitutes a rare disease; these numbers at first glance may seem insignificant. However, we must consider that there are more than 7,000 known rare diseases which affect approximately 25 million people. This is roughly translated to about 10% of the U.S. population.

Rare diseases are far more common than the term "rare disease" implies. Rare diseases are a significant contributor of chronic illness, disability, and premature death in both children and adults. They take a disproportionate share of our health care dollars and generate an enormous administrative burden on the healthcare industry as a whole because of their complexity and the expense of, in most cases, inadequate medical interventions. A Rare Disease Advisory Council (RDAC) is an advisory body providing a platform for the rare disease community to have a stronger voice in state government. RDACs address the needs of patients and families struggling with rare diseases by giving stakeholders an opportunity to make recommendations to state leaders on critical issues including the need for increased awareness, diagnostic tools and access to affordable treatments and cures.

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 RDACs, and the national trend is taking hold with rare disease stakeholders across the country diligently working to establish a mechanism to allow improved representation of the this historically underserved and often misunderstood population of people.

The symbol for rare disease awareness is a black and white striped 'Zebra' ribbon.

Rare disease advocates worldwide use a the zebra stripe ribbon because of the well-known medical expression.

The formation of the Nevada Rare Disease Advisory Council "the Council" was formed during the 2019 session of the Nevada Legislature. The council was assigned the following 13 specific duties:

1. Perform a statistical and qualitative examination of the incidence, causes and economic burden of rare diseases in Nevada.
2. Receive and consider reports and testimony concerning rare diseases from persons, the Division, community-based organizations, providers of health care and other local and national organizations whose work relates to rare diseases.
3. Increase awareness of the burden caused by rare diseases in Nevada.
4. Identify evidence-based strategies to prevent and control rare diseases.
5. Determine the effect of delayed or inappropriate treatment on the quality of life for patients suffering from rare diseases and the economy of Nevada.
6. Study the effect of early treatment for rare disease on the quality of life for patients suffering from rare diseases, the provision of services to such patients and reimbursement for such services.
7. Increase awareness among providers of health care of the symptoms of and care for patients with rare diseases.
8. Evaluate the systems for delivery of treatment for rare diseases in place in Nevada and develop recommendations to increase the survival rates and quality of life of patients with rare diseases.
9. Determine effective methods of collecting data concerning case of rare diseases in Nevada for the purpose of conducting epidemiological studies of rare diseases.
10. Establish a comprehensive plan for the management of rare diseases in Nevada, which must include recommendations for the state and local health authorities, public and private organizations, businesses and potential sources of funding, and update the comprehensive plan as necessary.
11. Develop a registry of rare diseases diagnosed in Nevada to determine the genetic and environmental factors that contribute to such rare diseases; and
12. Compile an annual report that includes a summary of the council's activities and any recommendations of the council for legislation or other policies.
13. A specialized license plate to support the Rare Disease Advisory Council and research and treatment for childhood cancer can be purchased through the Nevada DMV Specialized License Plates website.

The expression comes from an old saying used in teaching medical students about how to think logically regarding the process of differentiating between two or more conditions that share similar signs or symptoms. When you hear hoof-beats, think of horses, not zebras.

In the case of rare diseases- it is a zebra we are looking for and they are often extremely hard to identify in a world of horses. The zebra print ribbon seems to represent the difficulty people with rare diseases often face when seeking a diagnosis.

Our History

The NV-RDAC was formed under SB-315 during the 2019 session of the Nevada Legislature. The council was assigned several specific duties that range from performing a statistical and qualitative examination of the incidence, causes, and economic burden of rare disease in Nevada; including the development of a registry of rare diseases diagnosed in Nevada to determine the genetic and environmental factors that contribute to such rare diseases. As well as the responsibility of increasing awareness of the burden caused by rare disease; including improving awareness among health care providers of the symptoms and care required for patients with rare diseases.

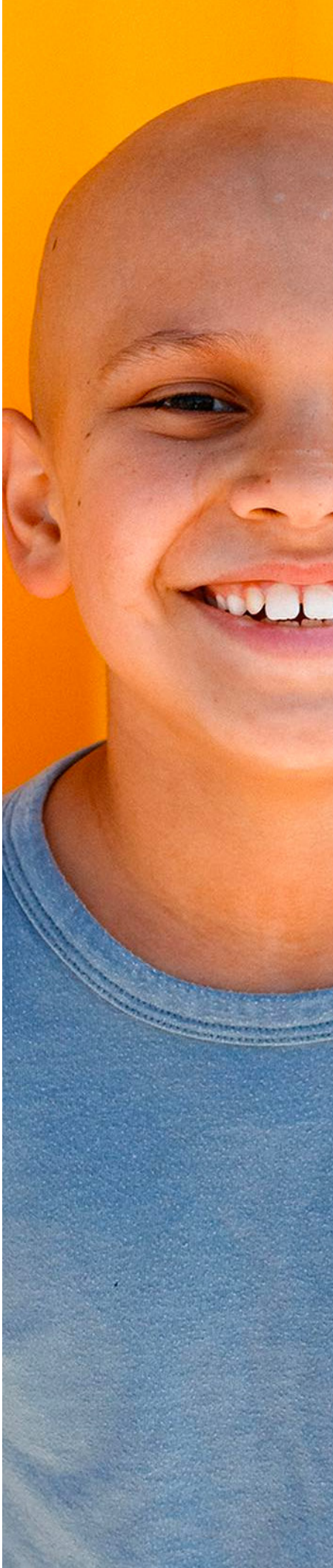
Unfortunately, as of yet, the state of Nevada has not provisioned their RDAC with the required resources to be effective in their efforts. We are hopeful that the Nevada DMV Specialized License Plate for childhood cancer initiatives will eventually grow to become a significant resource for the RDAC. For more information, please scan the QR code to the right to learn more about the license plate sales.

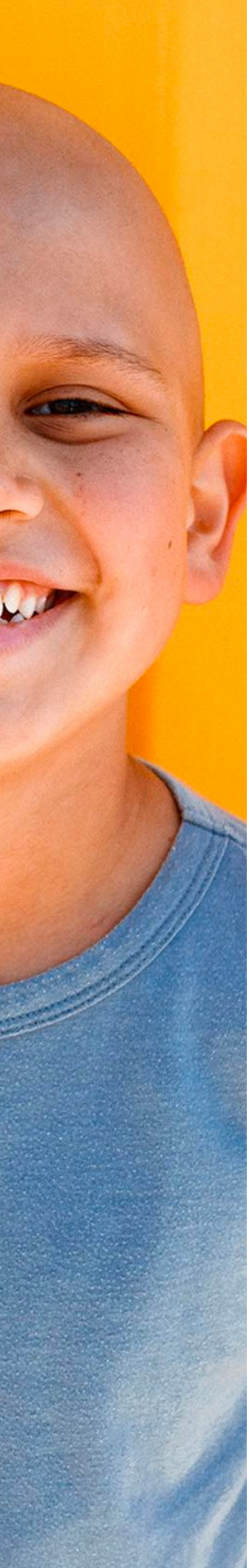
Scan to View
or click HERE

Total Fees

Numbering	Initial	Renewal
Sequential	$61	$30
Personalized	$96	$50

- Sequential plates are in stock at DMV offices.
- **Order personalized plates online**, or
- Use the **Application (SP 66)** (PDF) for in-person, mail or fax orders.





Even though a significant portion of the population has a rare disease, state policymakers typically lack in-depth knowledge about the rare disease community as a whole and the issues relevant to this community. This incomplete understanding contributes to common obstacles that rare disease patients face, such as delays in diagnosis, misdiagnosis, lack of treatment options, high out-of-pocket costs, and limited access to medical specialists.

Nevada was the 10th state to enact a Rare Disease Advisory Council. As of July 2022, there are 24 states with established RDACs and 9 more with pending legislation. The NV-RDAC was established to help bridge policymakers' gaps in knowledge, and serve as an advising body and liaison between the rare disease community and state government.

The council helps inform, evaluate, and offer recommendations on policies and issues relevant to the rare disease community. There are statutory requirements for NV-RDAC members to ensure there is a balanced representation of interested parties.

For example:

- Physicians and nurses who care for patients with rare diseases and hospital administrators that provide services to people with rare diseases, and employees of the Division of the Nevada Department of Health and Human Services
- Patients who have rare diseases and parents of children with rare diseases.
- Administrators of organizations from both northern and southern Nevada who provide services to patients suffering from rare diseases.

If you are interested in being considered for a future Council Member appointment to the RDAC, contact Ashlyn Torrez by phone 775-447-0263, by email at atorrez@health.nv.gov, or by mail at: 4150 Technology Way, Ste. 300, Carson City, NV 8970

Please submit a letter of interest and a resume or curriculum vitae. Materials submitted will be forwarded to the DHHS Director for consideration of possible appointment.

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or click HERE

NV-RDAC To Date:

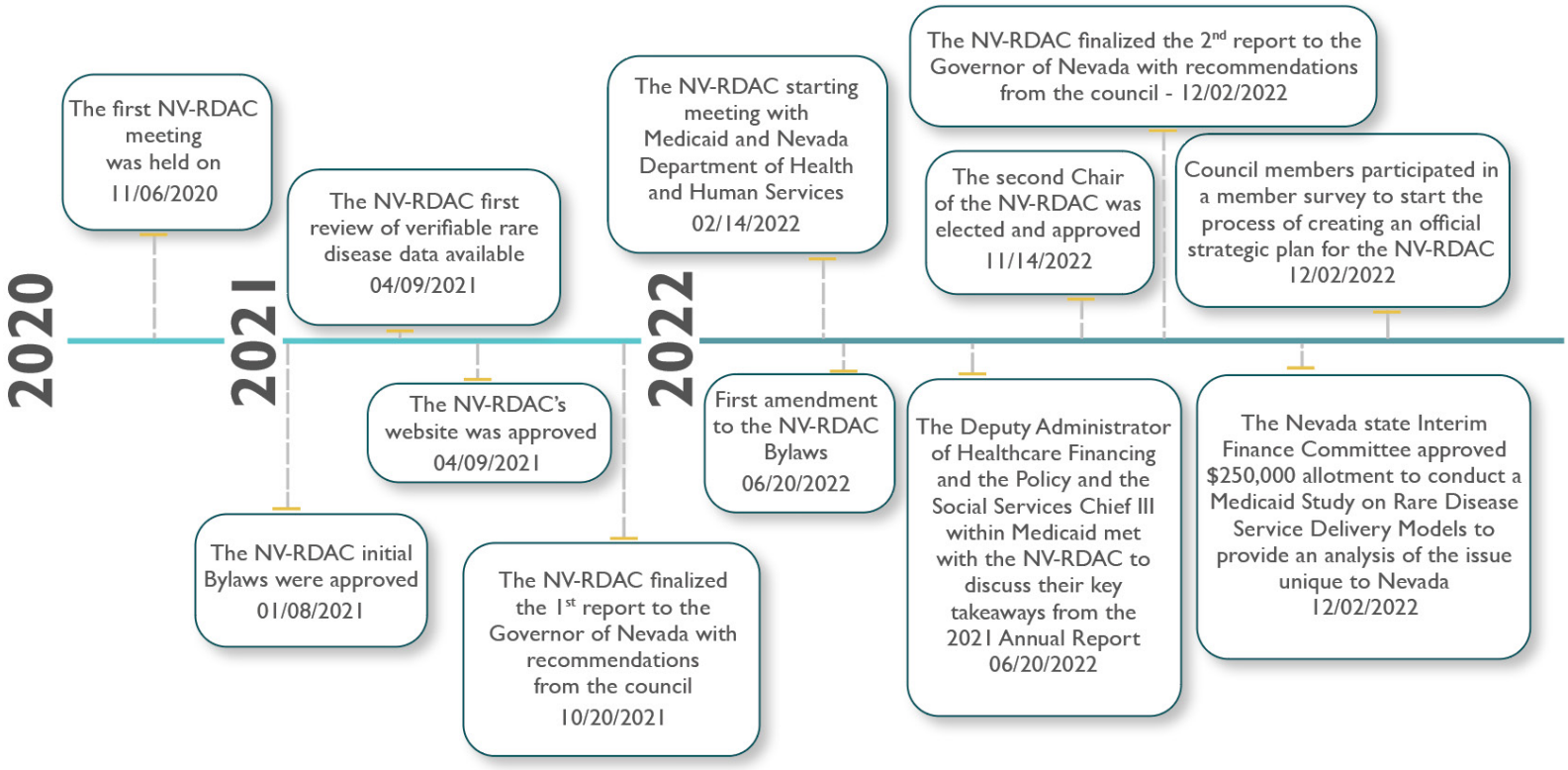
The Nevada Rare Disease Advisory Council has been tasked with several important deliverables, a few of them are as follows:

1. The development of a statewide registry of rare diseases diagnosed in Nevada to help the state better understand the prevalence of rare diseases as well as the burden of rare disease affecting the people of Nevada.

2. Evaluate the systems for delivery of treatment for rare diseases in place statewide and develop recommendations to increase the survival rates and quality of life of patients with rare diseases.

3. Establish a comprehensive plan for the management of rare diseases in Nevada, which must include recommendations for the state and local health authorities, public and private organizations, businesses, and potential sources of funding.

NV-RDAC Timeline



Current Rare Disease Areas of Focus

The NV-RDAC voted in December of 2020, that their initial focus would be on three primary categories of rare diseases:

1. All Childhood Cancers
2. Factor deficiencies and inherited platelet disorders
3. Newborn screening conditions- currently, the newborn screening in Nevada screens for 46 conditions

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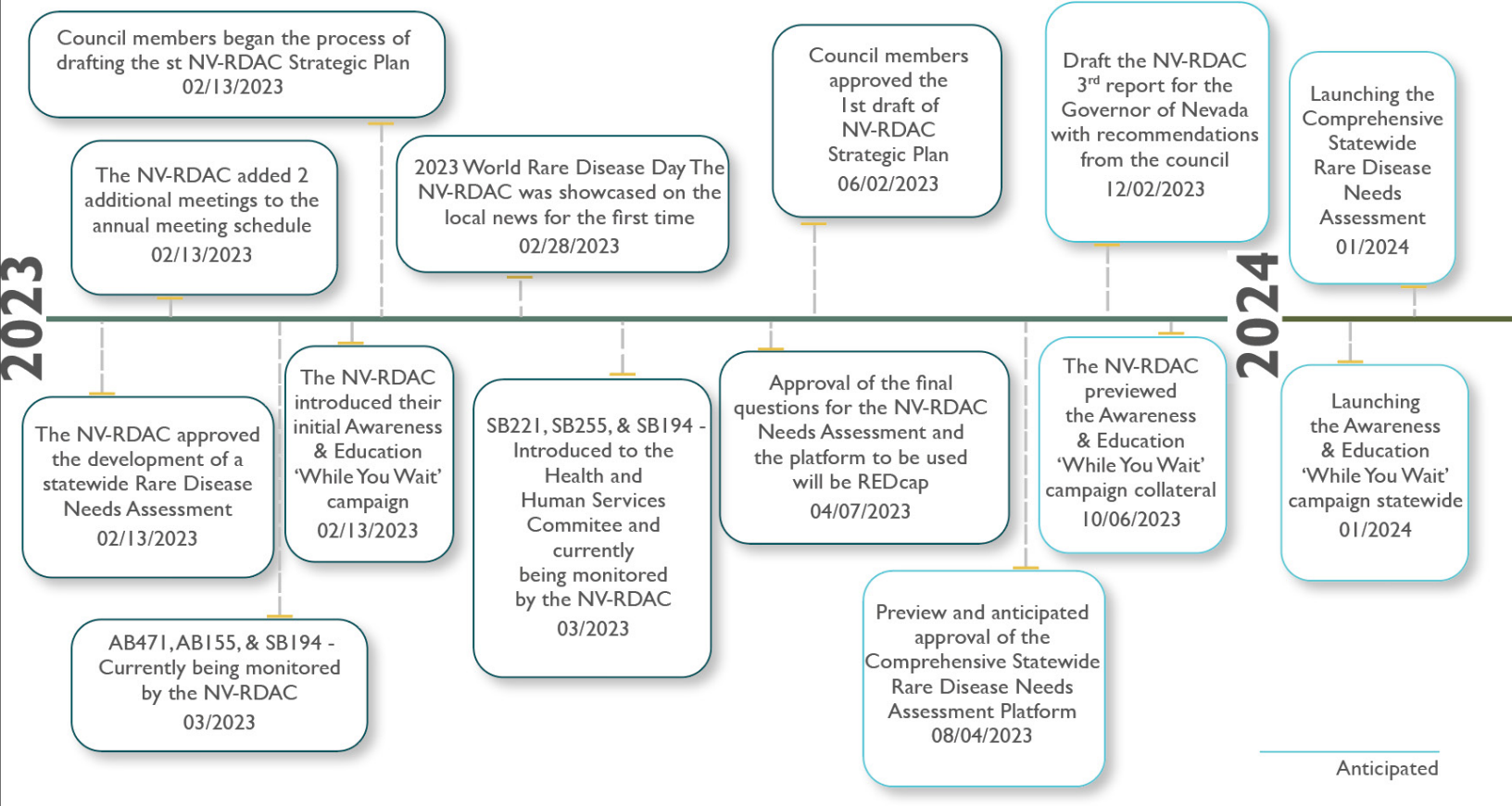


Preparing for this Strategic Plan

The Nevada Rare Disease Council members participated in an anonymous survey to gain perspectives on where their limited resources should be applied. The survey confirmed that the members of the NV-RDAC are not only engaged and willing to serve the state of Nevada on behalf of people with rare diseases, but they are also equally aligned in their personal and collective agendas to improve the overall landscape of rare disease management in Nevada. This includes access to high quality medical care, improving the knowledge base of the states healthcare providers, influencing positive change with payers who provide coverage for people with rare diseases, and educating the state's lawmakers to create new and improved regulations that positively impact people with rare diseases and the families who love them.

The survey instrument that the NV-RDAC council members used placed the 13 duties assigned to them into 6 different categories. The council members then ranked them in order of priority based on their current capabilities. The survey determined that the top priorities should be as follows, in the order of importance:

1) Create and Develop NV-RDAC's Awareness & Education Campaign
2) Leverage the network of Rare Disease Advisory Councils for Efficiencies and Effectiveness
3) Establish Continuity of Care for People Living with a Rare Disease in Nevada through the creation of a Comprehensive Statewide Rare Disease Plan
4) Address Health Equity and Disparities that Impact People with Rare Diseases
5) Develop a Nevada-Specific Data Collection & Analysis Platform
6) Launch a Comprehensive Statewide Rare Disease Needs Assessment

Scan the QR code to the right to view the summary of findings of the survey in the 02/13/2023 Meeting Minute attachments

Scan to View
or click HERE

Moving The Plan Forward

This strategic plan is the first statewide coordinated effort to address rare diseases in Nevada. Due to the great complexity of rare disease, the significant unmet needs, and the critical urgency associated with rare diseases, systematic reform is required.

While there are many rare diseases, they share countless commonalities. Informed by extensive stakeholder consultation through the 2022 council member survey, this strategic plan addresses this common ground. It represents the views of the rare disease sector and outlines the initial comprehensive, collaborative, and data-informed approach required to achieve the best possible health and well-being outcomes for people living with rare diseases in Nevada.

The NV-RDAC strategic plan is built upon the following 3 Pillars

- **Awareness and Education**
- **Care and Support**
- **Research and Data**

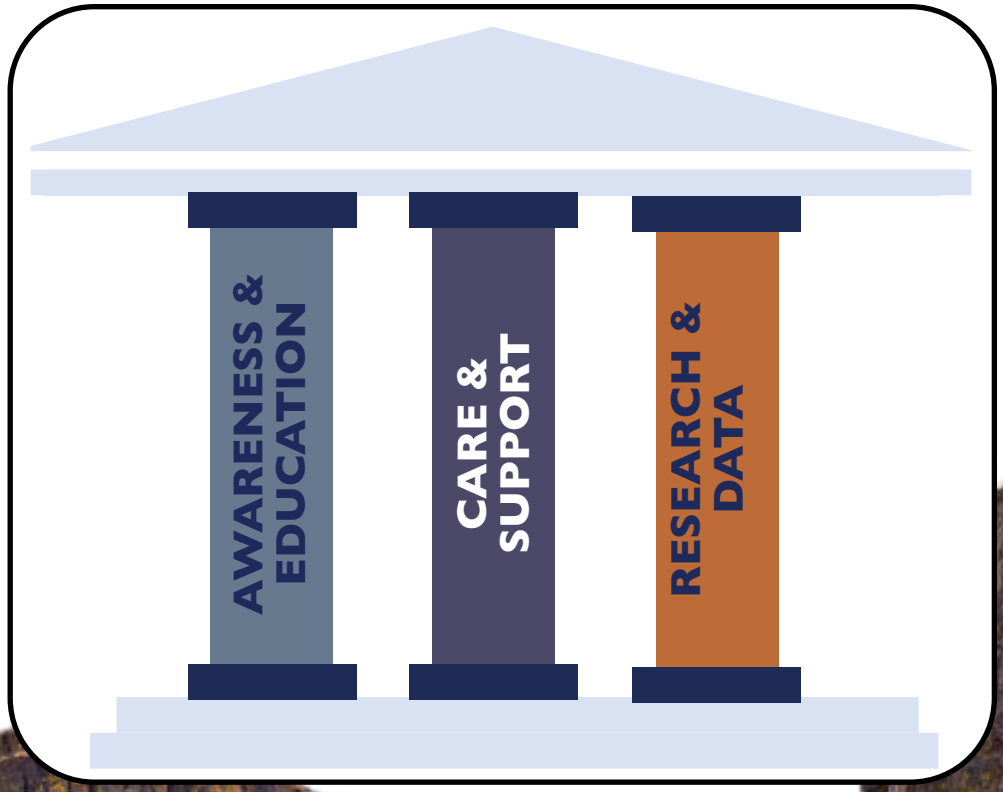
These pillars are easily recognizable to people living with rare diseases as well as the healthcare professionals providing services for them. For the purpose of this strategic plan, each pillar is presented seperately, yet is important to recognize that they are all interrelated and go hand-in-hand. As such, it is critical for us to address the priorities of each individual pillar while recognizing that reform and change to one pillar will create change and momentum in the others.

The importance of statewide leadership and coordination in rare diseases cannot be underestimated. It is going to require collaborative relationships, multi-stakeholder involvement and engagement, as well as shared governance and oversight.

This NV-RDAC strategic plan is critical in aligning the council's activities moving into the next three years. The council will continue to grow and evolve. We will be faced with new challenges and opportunities that will take priority. However, the pillars of this plan will remain the foundation of our collective efforts moving forward with clear goals and objectives.

NV-RDAC Pillars of Success

AWARENESS & EDUCATION	CARE & SUPPORT	RESEARCH & DATA
• Create and Develop NV-RDAC's Awareness & Education Campaign	• Establish Continuity of Care for People Living with a Rare Disease in Nevada through the creation of a Comprehensive Statewide Rare Disease Plan	• Develop a Nevada-Specific Data Collection & Analysis Platform
• Leverage the network of Rare Disease Advisory Councils for Efficiencies and Effectiveness	• Address Health Equity and Disparities that Impact People with Rare Diseases	• Launch a Comprehensive Statewide Rare Disease Needs Assessment



Our Approach to **Awareness & Education**

What are we hoping to achieve?

1. Increase every Nevadan's awareness of rare diseases and education on the resources available.

2. Ensure Nevadans living with a rare disease have access to information and education that empowers them to be active participants in their rare disease journey.

3. Develop a statewide rare disease workforce strategy that responds to current and future demands; including the impact of genomics.

Why is this Important?

Increased awareness and education at the individual and community level is important. It is common for people to be diagnosed with a rare disease that they and their family members never even knew existed. Due to the volume of rare diseases, it can be challenging to find a healthcare provider who is sufficiently experienced in the care of patients with rare diseases. Yet, the people diagnosed with a rare disease and the healthcare professionals providing the care are reliant on services and information for both the care and support of these diseases. Lack of awareness of rare diseases often contribute to people feeling isolated, misunderstood, and extremely frustrated. This lack of awareness also results in delays in diagnosis and treatment, potentially missing important opportunities for early intervention and improved outcomes [1].

While awareness is important, with over 7,000 known different rare diseases, it is impossible for any individual; including healthcare professionals to be aware of them all. Awareness activities must be supported by systematic identification, classification, and a prioritized response to rare diseases and undiagnosed rare diseases [2]. Therefore, it stands to reason, that people would not need to know everything about all rare diseases, but rather would need to know how to locate relevant information as it is needed. Rare disease data collection and use is further explored in Pillar 3.

Education about rare diseases needs to empower people living with a rare disease to become active participants in their rare disease journey. Education efforts need to appreciate the fact that people living with a rare disease are constantly learning, collaborating, and ultimately teaching each other, as well as healthcare professionals, about the nuances of these particular diseases.

There is also a serious need for concentrated education of the healthcare workforce that supports people living with rare diseases. We need to focus on increasing the healthcare industry's capacity to meet (and eventually exceed) the care and support requirements of people living with rare diseases. While this may apply to the entire healthcare workforce, there are certain segments of the workforce such as those providing mental health services, with known and urgent awareness and education needs.

Rare Disease, Genomics, and the Future...

We need to Get Ready!

The Diagnostic Odyssey- A search for answers that can last years and involve many different tests and consultations with clinicians across multiple specialties. Even if a diagnosis is reached, thereis often no treatment. Genomics can offer a long-awaited diagnosis that can bring some certainty an understanding where before there was none. Over 80% of rare diseases currently have a known genetic origin.

Fortunately, there is great opportunity to understand the cause of each rare disease and to provide an accurate diagnosis for each patient. Genomics plays an important role with a notable impact on public health. Many emerging technologies appear to play an essential role in diagnosis and for patients who remain undiagnosed. This future looks bright - as long as state policy can keep up with science.

Whole genome sequencing from a single blood test can pick up 31% more cases of rare genetic disorders than standard tests, shortening the expensive and frustrating "Diagnostic Odyssey" [3]. The faster, cheaper generation of genomic data is driving the integration of genomics into all healthcare specialties. It is likely that within the next decade, many health care professionals will be using genomics to diagnose and manage their patients- this is expected to be the case with rare diseases.

Nevada is not prepared for this genomic-based future. In order to keep up with scientific progress in the field of rare diseases, there are **three major actions** that we must take in Nevada:

1. Increased awareness of the state's actual incidence and burden of rare diseases
2. Accessible and equitable genetic testing for all Nevadans (when appropriate) recognizing that new genomic technologies are transforming healthcare.
3. State policy that requires Medicaid and commercial insurance coverage for genetic testing; particularly for children - as 70% of rare disease start in childhood.

Increased awareness of various rare diseases would help clinicians quickly recognize symptoms and pursue a diagnosis. Free or low-cost genetic tests would eliminate the need to rule out multiple conditions and get a quicker diagnosis. This could allow patients to access medications or investigational therapies, and hopefully, slow or halt disease progression. Insurance coverage of genetic testing would help patients access potentially life-saving therapies sooner.

Our Priorities and Actions
Awareness & Education

Priority One: **Create and Develop NV-RDAC's Awareness & Education Campaign**

Actions:

1. To create and launch our 'While You Wait' campaign (in English and Spanish). This campaign is a suite of collateral materials to raise awareness of the NV-RDAC to be sent out to all relevant provider offices statewide. The intention is to provide awareness and educational materials to a variety of Nevadans while they are waiting in their healthcare provider's office. This material will be available for a wide variety of audiences and appropriate for a range of specialties. This also allows us to seek participants for the Rare Disease Needs Assessment, which will link Pillars 1 and 3.

2. Create a Media Campaign using a variety of social media channels (Facebook, Instagram, and Linkedin) as well as an in-kind/sponsored traditional news and print campaign (magazines and newspaper) to bring awareness of the NV-RDAC, the important work we are doing, and how to support the NV-RDAC initiatives.

3. Develop an awareness and education program that empowers frontline healthcare professionals to provide culturally safe and appropriate medical care to support all Nevadans living with a rare disease. While also identifying existing gaps in the workforce that provide medical services to people living with rare diseases and outline a path towards a sustainable workforce, taking into account the impact of genomics.

4. Expand the capabilities of the NV-RDAC website. In order to develop and promote the use of an easily accessable multi-purpose digital repository for both healthcare professionals and people living with rare diseases.
    - Healthcare professionals: In order to support these healthcare professionals to consider a rare disease diagnosis when people present with unexplainable symptoms.
    - People with rare diseases: In order to support them and their families to identify, not only appropriate info but also who are the professionals in the state of Nevada who can treat them.
5. Promote through education and awareness efforts the specific needs of people with a life-limiting rare disease and the importance of providing thoughtful and appropriate end-of-life care and support services.

Priority Two: **Leverage the Network of Rare Disease Advisory Councils for Efficiencies and Effectiveness**

Actions:

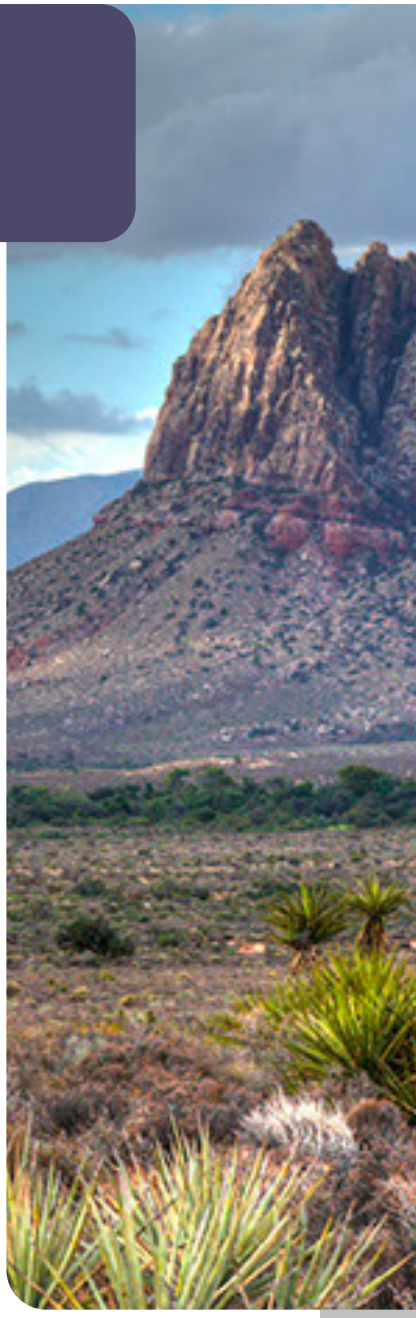
1. Ensure collaboration and consultation occurs between Nevada state policy & lawmakers with rare disease stakeholders including education providers, healthcare providers, payers, and community-based nonprofit organizations.

2. Capitalize on the existing multi-purpose digital resources already available nationally and internationally in order to bridge the available information on the NV-RDAC website. Ensuring the ease of access of information to both the people living with rare diseases as well as the healthcare workforce providing them with medical services.

3. Build on existing activities of local and national rare disease organizations to raise awareness of care and support services available to people living with rare diseases in Nevada and identify gaps and opportunities for improvement.

4. Strengthen the connection with state lawmakers and both the local rare disease community and the healthcare professionals providing care to them. Improve communication on a range of issues that people with rare diseases face in Nevada.

2 Our Approach to Care & Support



What are we hoping to achieve?

1. Influence improved care and support that is integrated and appropriate for all Nevadans living with a rare disease.

2. Set the standard that diagnosis of a rare disease in Nevada is timely and accurate while ensuring Nevadans have equitable access to qualified healthcare and the best treatment options.

3. Create awareness and educational opportunities that inspire integration of mental health, and social & emotional well-being into rare disease care and support.

Why is this Important?

Early diagnosis enables the best clinical care, treatment options, access to services, peer support, increased reproductive confidence and access to participation in clinical trials. Yet, diagnostic delay and misdiagnosis are common in rare diseases.

It has been reported that at large number of people living with a rare disease worldwide are impacted by a diagnostic delay of more than five years, while almost half have received at least one misdiagnosis [4]. Currently, Nevada-specific statistics in diagnostic delay and misdiagnosis are unknown. It is believed by the members of the NV-RDAC that it is highly likely that Nevadans experience the same delay and misdiagnosis as seen around the world. Rare diseases data collection and use is explored further in Pillar 3.

Significant delays in diagnosis as well as experiencing misdiagnosis have physical, psychological, emotional and financial costs for the person and family living with a rare disease. There is a severe need for rare disease care and support to be less fragmented and more integrated. The medical care and community support required for people living with a rare disease needs to be both personal and family-centered. People with a rare disease often require large interdisciplinary teams of doctors, nurses and allied health professionals who work in different settings (primary care, hospital, emergency departments, community allied health) to manage the multiple medical problems and disabilities experienced. Their health needs can change throughout the course of their lifetime, and these changes can be rapid and critical. Additionally, they often have complex support needs that extend beyond healthcare that include disability, social/welfare, mental health, education,

employment and housing. It is not just the person living with a rare disease who needs support. Their family and the healthcare professionals also have high and significant support needs.

Rare disease care and support should meet the needs of patients and the healthcare professionals while taking into account local contextual factors, such as existing services and structures; resources (including funding and workforce), workforce expertise, and the preferences of people living with a rare disease and their families and providers.

Living with a rare disease does not only affect a person's health; it impacts every facet of their life, including education, employment, as well as mental & physical health. For example, it is currently difficult for people living with a rare disease to navigate their way through the health and disability systems as there is a lack of clear referral pathways and clinical coordination.

Valuable care and support are provided by tax-exempt, rare disease organizations both nationally and locally. This includes peer support, the provision of information, access to resources, and individual and systemic advocacy. Many rare disease organizations are run by people living with their own rare disease challenges, which can leave them vulnerable and affect their ability to offer services over the longrun (including awareness, education, care, support and research). The state of Nevada has an opportunity to work directly with the NV-RDAC to identify solutions that could result in the stabilization of this fragile yet critically important support system, that people living with a rare diseases access.

There are limited treatment options for rare diseases, and even when a treatment does exist, financial support may not be available in Nevada and thus accessibility may be limited. Reimbursement of health technologies for rare diseases, using models designed for more common diseases, is challenging as smaller patient numbers impact cost effectiveness, and there is often less clinical evidence available due to the challenges of conducting large-scale clinical trials. This highlights the importance of alternative approaches to both identifying and funding the best possible treatment options.

There are many examples of an approved medicine (for a more common condition) that demonstrate benefits for rare diseases. However, due to small numbers, it is not always commercially viable for companies to seek reimbursement for a rare disease indication. Without government reimbursement, many rare disease medicines are unaffordable. As many rare diseases are progressive, time is often critical, making timely and equitable reimbursement essential for people living with a rare disease so that they may benefit from new and transformative treatment options.

Our Priorities and Actions
Care & Support

Priority One: **Establish Continuity of Care for People Living with a Rare Disease in Nevada through the Creation of a Comprehensive Statewide Rare Disease Plan**

NV-RDAC is focused on establishing a comprehensive statewide strategy to deliver safe, sustainable, world-class medical care for people living with a rare disease in Nevada. The goal is for all Nevadans to receive the correct diagnosis and the most current and medically appropriate treatment provided by a team of highly qualified and skilled professionals. All while ensuring access (if appropriate) to participation in any rare disease clinical trial, for which they are eligible.

Actions:

1. Educate Nevada lawmakers on the importance of building a broad range of care and support services that are responsive to the unique needs of people living with a rare disease.

2. Influence Medicaid and commercial payers in Nevada to use published best practices for the standardization of available care and support for people living with rare diseases. Ensuring these standards of care and support are integrated throughout the entire healthcare delivery system.

3. Work to reduce fragmented care, ensuring policy addresses the full range of needs of people living in Nevada with a rare disease; including healthcare, disability, housing and education.

4. Increase awareness among healthcare professionals, payers (Medicaid and commercial), as well as rare disease community-based organizations of the importance of palliative care in rare disease management. Palliative care can achieve a wide range of objectives for people and their families living with a rare disease; improving both the quality of life and thoughtful and appropriate end-of-life care.

The Right **Diagnosis**
+ The Right **Treatment**
+ The Right **Team**
+ The Right **Environment** = **CONTINUITY OF CARE**

Priority Two: **Address Health Equity and Disparities that Impact People with Rare Diseases**

80% of rare diseases are genetically based, yet access to genetic testing and counseling is extremely limited and often completely unavailable to people with limited resources. The development of a statewide standard of practice that ensures the timely access to appropriate genetic testing and genetic counseling, will result in healthcare professionals being able to quickly deliver high-quality care to many people with rare diseases.

The Diagnostic Odyssey which translates into the relentless persuit of diagnostic answers can take several years and often involves many different tests through consultations with clinicians across multiple specialties. This historically accepted practice is not only incredibly frustrating to people seeking diagnosis with rare diseases and the healthcare professionals that treat them, it is also unnecessary, extremely expensive, and leads to chronic financial toxicity.

Financial toxicity in healthcare, particularly in the rare disease sector has become a serious, national topic and Nevada has not escaped the conversation. There is a common misconception that financial toxicity is simply the high cost of care, but it's more than that. Most families are pushed to the edge and often beyond their financial limits, and this still does not convey the full expenses that result from living with a rare disease.

Actions:

1. Influence the state of Nevada's policy to adequetely support people living with a rare disease to have timely and equitable access to both diagnostic opportunities as well as the most current scientifically-proven treatment options for their rare disease.

2. Work with lawmakers in the state of Nevada and payers including Medicaid and commercial to build a equitable and timely delivery of medically-appropriate and scientifically-proven services such as genetic testing (diagnostics) and gene therapies (treatments) as well as genetic counseling to Nevadans who are suspected of having a rare disease or those with an increased chance.

3. Encourage the state of Nevada to develop policy that ensures equitable access to appropiate genetic diagnostic testing, genetic counseling, and the resulting healthcare interventions for all Nevadans living with a rare disease or seeking answers for undiagnosed medical conditions.

4. Identify the statewide specific roots of financial toxicity for patients and families living with rare diseases in Nevada.

The single most important driver for the right diagnosis, right treatment, right team, and right environment **is defining and setting statewide standards of care** to achieve the very best clinical outcomes, while putting the needs of the patient and their families at the center of all decisions.

3 Our Approach to Research & Data

What are we hoping to achieve?

1. Establish coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases in Nevada, informing policymakers, payers, and healthcare professionals statewide.

2. Develop a statewide research strategy for rare diseases to foster, support, and drive all types of research for rare diseases while systematically addressing the identified gaps in care and support.

3. Influence the state of Nevada to recognize the unique needs of patients living with rare diseases using research and data to educate our lawmakers on the level of reform that is required for our state to adequetely ensure Nevadans have equitable access to qualified healthcare and the best treatment options.

Why is this Important?

In Nevada, data for most rare diseases is not captured in either health information systems or registries and there is no coordinated strategy to collect, measure, build and translate the data that does exist. The NV-RDAC has identified the need for a statewide, coordinated, and systematic approach to the collection and use of rare disease data, including registries. Such an approach will enable monitoring and the accumulation of knowledge about rare diseases to inform clinical practice, research and health policy planning.

For key decision-makers at all levels, greater knowledge of rare diseases can facilitate more responsive and appropriate services for people living with a rare disease. The NV-RDAC is eager to lead significant positive change in this space.

For many rare diseases, there are a number of barriers to effective research and often no active research programs at all. One of the biggest challenges is that rare diseases at the individual disease level have small patient numbers and are often very complex. When looking at rare diseases collectively, we can learn alot on how to improve the care and support that people living with rare diseases need. This is further explored in Pillar 2. Depending on the specific rare disease, research priorities can be different. For example, while funding for translational research may be important for some, other rare diseases are not yet in the position to be prioritized in translational research leaving unmet needs for basic discovery initiatives or investment into data collection and genomics.

For many people living with a rare disease, participation in a clinical trial may be the only way to access treatment. Estimates are as high as 90% of people living with a rare disease are interested in joining a patient registry, in recognition of the key role that registries play in linking people living with a rare disease with clinical trials for new treatment options [5].

The translation of rare disease research into clinical settings, while currently hampered, is vital. This two-way relationship benefits from active participation by patients, their families and their healthcare providers, and patient advocacy groups to ensure the best possible outcomes for people living with a rare disease.

There is an understanding in the rare disease community that, while research may not lead to better outcomes for people currently living with a rare disease, participation in research may drive change for future generations. This is supported by outcomes of the Rare Barometer survey undertaken in February 2018 by EURORDIS, Rare Diseases Europe [6].

Research into rare diseases must address existing gaps and the coordination of research projects must be prioritized. Improving policy settings and statewide collaborations, will help to drive strong research and innovation for all rare diseases in Nevada. Research into rare diseases needs to inform evidence-based policy across all systems, extending beyond healthcare to incorporate disability, social/welfare, mental health, education, employment and housing.

3 Our Priorities and Actions
Research & Data

Priority One: **Develop a Nevada-Specific Data Collection & Analysis Platform**

There is an urgent need for the expansion of rare disease expertise and further development of evidence-based rare disease care. Limited data is a common feature in rare diseases. This is heightened by poor quality, disjointed collection methods and the ineffective use of data for rare diseases. Such limitations are evident across a range of areas, from health system classification to research. Research, monitoring, and ongoing evaluation are critical in rare diseases because, ultimately, if we are not counting rare diseases, people living with rare diseases do not count.

Actions:

1. Establish a statewide community of stakeholders focused on rare disease data collection and research.

2. Improve rare disease data collection efforts to include a statewide approach to collecting data on the 3 focus groups of rare diseases; childhood cancer, bleeding, and newborn screening.

3. Influence the state to undertake broad epidemiological surveillance of rare disease to support decision makers and lawmakers to access the information they need to inform the health and well-being of Nevada's living with rare disease.

4. Develop a statewide registry for rare diseases to foster, support and drive all types of research for rare disease.

5. Support collaborative research into rare disease in Nevada (and nationally) by investigating and promoting options that enable Nevadan's living with a rare disease to participate in clinical trials and other research activities.

Priority Two: **Launch a Comprehensive Statewide Rare Disease Needs Assessment**

The NV-RDAC is seeking survey participants to gain further information from individuals and their families and caretakers living with rare disease in the state of Nevada. The primary objective of this needs assessment is to gather insights and perspectives directly from Nevada residents to increase our understanding on the barriers to care. The findings of the needs assessment will advise on quality of care, or lack thereof, educational needs, diagnosis, and other factors related to living with a rare disease. We are anticipating on collecting data from the needs assessment for 24 months starting in January of 2024.

Actions:

1. Research our peer groups and existing RDACs across the country to develop a series of appropriate questions for our needs assessment.

2. Build, through REDCap, the needs assessment survey instrument. The needs assessment is a collection of questions aimed toward data collections focused on gaps in care and opportunities for statewide improvement. Through this data collection, we are intertwining initiatives in Pillars 3 and Pillar 1.

3. Launching the first comprehensive needs assessment survey to be available in provider offices as well as other available platforms statewide. Anticipated launch date is January 2024.

4. Showcasing our findings on an annual basis through our required NR-RDAC Annual Report to the Governor's office.

5. Champion transformative healthcare policy development through publishing our findings for public review. While also ensuring all Nevada lawmakers, Nevada Medicaid leadership, and relevant commercial payers understand how to use this information to not only improve the lives of Nevadans living with rare diseases but to also understand and address the health inequities and disparities that impact this patient population.

Summary & Call to Action

We face many challenges in the state of Nevada when it comes to the fight against rare diseases. One of the more significant challenges is the lack of statewide coordination. There are many different organizations and individuals supporting those living with a rare disease in the state. Even with all the effort and significant work being done for them, there is essentially little coordinated effort between southern and northern Nevada. Working together as a united front would allow us to harness our collective efforts and capitalize on our combined strengths and resources to bring a significant amount of influence to our state leaders and lawmakers to help us advance the fight against these terrible diseases.

This strategic plan is our call to action. In this document, we have taken you through the NV-RDAC's current initiatives and outlined our focus through 2025. This document provides an outline of what we feel are the minimum requirements to bring the state of Nevada to acceptable standards for the care and support of the people living with rare diseases.

Our plea to the leaders and lawmakers of Nevada is to support a meaningful statewide action plan that addresses the unique needs of people living with rare diseases, thier families, and the heathcare professionals that care for them. A coordinated effort and statewide strategy is the only way for Nevada to influence the type of policy change required to meet the needs of the people living with a rare disease in our state.

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