# NEVADA RARE DISEASE ADVISORY COUNCIL

**DRAFT MEETING MINUTES**
**Date: February 07 2025**
**9:32 AM – 11:09 AM**

Meeting Locations:
Pursuant to NRS 241.020(3)(a) as amended by Assembly Bill 253 of the 81st Legislative Session, this meeting was convened using a remote technology system and there was no physical location for this meeting.
Chair Annette Logan-Parker opened the meeting at 9:32 am.

1) INTRODUCTIONS AND ROLL CALL

**COUNCIL MEMBERS PRESENT:**
Annette Logan-Parker (CHAIR); Ihsan Azzam, MD, PhD; Jennifer Millet, DNP, RN; Valerie Porter, DNP, APRN, AG-ACNP-BC, MBA; Naja Bagner; Paul Niedermeyer; Christina Thielst, LFACHE, MHA; Sumit Gupta, MD; Brigette Cole; Kim Anderson-Mackey; Melissa Bart-Plange; and Verena Samara, MD (Quorum=9)

**COUNCIL MEMBERS ABSENT:**
Gina Glass (Vice-Chair); Amber Federizo, DNP, APRN, FNPBC; Craig Vincze, PhD (excused); Pamela White (excused); and Devraj Chavda, MD

**DIVISION OF PUBLIC & BEHAVIORAL HEALTH (DPBH) STAFF PRESENT:**
Ashlyn Torrez, *Health Program Specialist I, Office of State Epidemiology (OSE), DPBH*; Kevin Dodson, *Administrative Assistant III, OSE, DPBH;* Kagan Griffin, MPH, RD, *Operations Manager, OSE, DPBH,;* and Jeri Borges *Administrative Assistant III, OSE, DPBH*

**OTHERS PRESENT:**
Esther Y Kwon; Jen L. Thompson, *Health Program Manager II, Office of Analytics (OoA), Department of Health and Human Services (DHHS);* Linda Anderson; Kem Wong, *Cancer Registrar, Cure 4 The Kids*; Po-Ju Wu, *Contract Biostatistician, OoA, DHHS;* Stefanie Abraham; and Dr. Mark Nunes, *Cure 4 The Kids*

Roll call was taken and is reflected above. It was determined that a quorum of the Rare Disease Advisory Council (RDAC, the Council) was present.

2) PUBLIC COMMENT:

Chair Logan-Parker opened the floor for public comment.

Hearing none, Chair Logan-Parker moved on to the next agenda item.

3) FOR POSSIBLE ACTION: Approval of minutes from December 6, 2024, Rare Disease Advisory Council Meeting. – *Council Members*

Chair Logan-Parker stated the minutes have been posted on the Department of Health and Human Services (DHHS) website and asked the Council for a motion to approve the minutes.

**Council member Jennifer Millet motioned to approve the meeting minutes from prior council meeting dated 12/06/2024. Council member Kim Anderson-Mackey seconded the motion to approve. There were no objections. A quorum voted to approve the prior meeting minutes.**

6) INFORMATIONAL ITEM: Presentation of the data dashboard for sickle cell disease (SCD) and systemic erythematosus lupus (SLE). – *Jen L. Thompson, Health Program Manager II, Office of Analytics, Director's Office, Department of Health and Human Services (DHHS); and Ashlyn Torrez, Health Program Specialist I, Office of State Epidemiology (OSE), DPBH, DHHS*

Chair Logan-Parker introduced Jen L. Thompson with the Department of Health and Human Services (DHHS) and opened the floor to Jen L. Thompson.

Jen L. Thompson introduced herself and explained that the data dashboard covered not only sickle cell and lupus but other rare diseases as well. Her team collected hospital billing data, vital records, and newborn screening data from the Nevada State Public Health Lab. Using ICD-10 codes, individuals with hospital visits related to lupus or sickle cell disease were identified, even if the visits were not specifically for those conditions. The data was displayed on the dashboard is similar to the COVID response analytics, with a current status tab for easy tracking. It included five years of hospital billing and visit records, newborn screenings, and death records. Hospital visits were categorized by lupus and sickle cell disease, while newborn screenings identified both the disease and sickle cell trait at birth. Death records captured cases where lupus or sickle cell were contributing factors, with trends over time showing changes across years. Month-by-month views of hospital visits and newborn screenings, along with demographic details such as age group, race/ethnicity, and gender, were provided. Despite some demographic gaps, efforts ensured accuracy. Plans included adding childhood cancers and other rare diseases, with input needed on prioritization.

Chair Logan-Parker asked whether all provider data in the community would be reported through the new reporting mechanism, currently referred to as "hospital." She also inquired if the focus was solely on data received from actual inpatient facilities.

Ms. Thompson replied she did include inpatient admissions, emergency department encounters, ambulatory and outpatient surgery, and then other outpatient facilities.

Chair Logan-Parker opened the floor to questions.

Council member Dr. Ihsan Azzam praised the dashboard and asked if there would be prioritization of diseases.

Ms. Thompson replied that the Council would be asked to identify the top diseases to prioritize.

Chair Logan-Parker added that providing a list of prioritized diseases would be possible.

Council member Kim Anderson Mackey asked about the lag time for processing the data.

Ms. Thompson replied that data was currently processed yearly, but quarterly processing was possible.

Dr. Ihsan Azzam commented that quarterly processing would be ideal.

Ms. Thompson noted that cancer data could only be processed yearly.

Chair Logan-Parker mentioned that Cure 4 The Kids had hired a cancer registrar, which was expected to improve cancer data for the state of Nevada.

Kem Wong, while reviewing the hospital website, asked how the data was organized.

Ms. Thompson responded that the data was organized by discharge date for hospital visits.

Chair Logan-Parker added that an operational meeting was scheduled to address data-related details like that.

Dr. Mark Nunes asked about any crossover or potential to integrate data with the 10 birth defects that the CDC tracks.

Ms. Thompson answers that it is something that she can include.

Chair Logan-Parker asked if the dashboard would be publicly accessible, how stakeholders could interact with the data, and whether a data-sharing agreement would be required.

Ms. Thompson stated that stakeholders would need a data-sharing agreement to interact with the data. All dashboards, once finalized through the review process, were to be made available on the Office of Analytics website through the portal.

Chair Logan-Parker also asked how Ms. Thompson preferred the RDAC to provide her team with recommended additional diagnoses for the dashboard.

Ms. Thompson replied that collaboration with Ashlyn and her team was preferred.

Council member Paul Niedermeyer asked if anyone had ever attempted to run a query in Epic or Cerner with some of the larger hospitals to determine the number of rare diseases occurring, potentially providing that data at a county level.

Ms. Thompson replied that no such queries had been conducted within their office. She noted that this was a new area for the Office of Analytics to assist with but indicated that efforts could be made to begin reviewing and identifying such data.

Dr. Azzam asked if Ms. Thompson obtained data from the death registry through death certificates.

Ms. Thompson confirmed that the data was sourced from there.

Dr. Azzam expressed interest in reviewing data on adult-onset diseases.

Chair Logan-Parker mentioned the payer portal and its potential to improve certain data collection processes.

Ms. Thompson explained that insurance providers, including Medicaid and Medicare, were required to report de-identified data to the all-payer claims database, which had recently launched. The database was set to collect data in three categories: prescriptions, medical services, and potentially dental care. Usable data was expected by fall 2026, after processing and cleaning by the state's vendor.

8) INFORMATIONAL ITEM: Introduction and Question and Answers (Q&A) with Dr. Mark Nunes – *Chair Annette Logan-Parker; and Dr. Mark Nunes, Director of Genetics, and Metabolism, Cure 4 the kids Foundation*

Chair Logan Parker introduced Dr. Mark Nunes.

Dr. Nunes shared his background, recalling a lecture by Anthony Fauci during medical school that provided an unexpectedly high level of education. He completed a pediatric residency with the Air Force in Northern California, followed by enrollment in an adult genetics program at the University of Washington instead of the intended pediatric program. After completing a fellowship, he managed a Deoxyribonucleic acid diagnostic lab for the Air Force in Biloxi, Mississippi, for seven years, handling cases from prenatal diagnosis to Alzheimer's disease. Post-military service, a child abuse fellowship was pursued before co-directing a metabolic bone program at Ohio State University. This led to directing the genetics program at Kaiser San Diego while serving on the UC San Diego faculty. Over the past decade, efforts focused on advancing technologies for early genetic disorder diagnoses, shifting from infant identification to second-trimester detection, with the current decade emphasizing rare disease treatment and expanded access to therapies for affected children and families.

Chair Logan-Parker asked Dr. Nunes to share significant challenges encountered in his career related to rare diseases and genetics.

Dr. Nunes stated his career was spent in closed healthcare systems, including the military and Kaiser, where access to genetic services depended on appropriate insurance coverage. In other healthcare settings, access to new genetic technologies posed significant challenges, particularly for patients in need. Whole exome sequencing, for example, was more accessible to patients in affluent areas, while those from underserved communities often faced little to no access. Financial barriers and insurance approvals frequently determined whether patients could receive advanced genetic testing. Additionally, genetic discrimination remained a recurring challenge, emphasizing the need for a strong consenting process for families undergoing genetic testing.

Chair Logan-Parker asked about the most pressing gaps in access to genetic services in Nevada.

Dr. Nunes responded by highlighting the insufficient number of genetic providers, with only a few geneticists in Las Vegas who had limited patient access and far fewer genetic counselors than needed. The recommended ratio of one genetic counselor per 100,000 people and one geneticist per 250,000 was not being met, leaving a significant gap in care. Despite strong technical capabilities and access to next-

generation sequencing in Nevada, the lack of practitioners to interpret and communicate genetic data to patients and families remained a major challenge.

Chair Logan-Parker asked Dr. Nunes to share his thoughts on the importance of licensure for genetic counselors and its potential impact on Nevada if made mandatory.

Dr. Nunes explained that genetic counselor licensure, a national movement led by the National Society of Genetic Counselors, allows counselors to practice independently without physician oversight. The absence of licensure in Nevada permitted anyone to provide genetic counseling, resulting in unregulated services lacking the specialized expertise of certified genetic counselors. While physicians and specialists offered genetic counseling, their training was limited compared to certified professionals, making licensure essential for improving access and quality of genetic services.

Chair Logan-Parker then asked how the RDAC could support this initiative.

Dr. Nunes responded that since December 2nd, efforts had aligned to identify genetic counselors in Las Vegas and Reno and to support the push for licensure, ensuring families had access to certified professionals for discussing genetic conditions, reproductive options, and emerging treatments for rare genetic diagnoses.

Chair Logan-Parker asked Dr. Nunes to explain the significance of whole genome sequencing and whole exome sequencing in diagnosing rare diseases, particularly for critically ill newborns.

Dr. Nunes highlighted that whole exome sequencing, first described in 2009, had advanced over the past 15 years to allow simultaneous testing of multiple genes. This technology enabled the diagnosis of rare diseases through targeted panels for conditions such as hearing loss and aortic dissection. Whole exome sequencing, focusing on the expressed regions of the genome (about 1% of DNA), significantly improved diagnostic yields for conditions like autism, increasing detection rates from 10-15% to 40-50%. Whole genome sequencing, examining 100% of the genome, emerged as a powerful tool in the past five to seven years, providing rapid results in critically ill newborns, sometimes within 24 hours, though the typical turnaround ranged from five to seven days. About 30 newborns annually in Nevada would meet the criteria for this testing, allowing early diagnosis, treatment adjustments, and avoidance of unnecessary tests. In cases of fatal genetic conditions, early diagnosis offered families critical information for timely palliative care.

Chair Logan-Parker then asked for examples from other states that Nevada could learn from regarding newborn screening.

Dr. Nunes cited Israel's national project, which offered rapid whole genome sequencing across all hospitals and published its findings in *Journal of the American Medical Association Pediatric* in 2024. While states like California and New York reimbursed rapid whole genome sequencing, others, including Nevada and Texas, did not. In 2021, the American College of Medical Genetics recommended whole exome or whole genome sequencing as the first-line test for individuals with intellectual disabilities, autism, or multiple congenital anomalies, surpassing the 10-15% diagnostic yield of cytogenetic microarray with a 40-50% success rate through whole exome sequencing.

Chair Logan-Parker asked Dr. Nunes to discuss immediate actions or initiatives recommended for the RDAC to prioritize in improving care for individuals with rare diseases in the state.

Dr. Nunes emphasized that funding proposals for genetic testing needed to highlight the importance of personnel infrastructure to relay and discuss results with families. Any initiative to expand state-funded genetic testing had to include a qualified provider network to assist families in navigating their results. Establishing a strong genetic community in Nevada was deemed essential for supporting families with rare diseases and improving access to genetic services.

Chair Logan-Parker asked Dr. Nunes about opportunities for Nevada to position itself as a leader in rare disease research and treatment.

Dr. Nunes responded that Nevada's relatively small population and annual birth rate made scaling up sequencing efforts for newborn screening a feasible possibility. Implementing DNA-based tests alongside current analytes could introduce precision medicine components for hundreds of disorders, with sequencing capacity already available in the state. Unlike larger states such as California and New York, Nevada's size allowed for flexibility and innovation in genetic testing infrastructure. The state also had the potential to serve as a model for other regions, including countries with dispersed rural populations, by demonstrating how to provide equitable access to advanced genetic technologies.

Chair Logan-Parker asked one final question regarding the single most important action Nevada could take to improve genetic services for the rare disease population.

Dr. Mark Nunes responded that building a strong genetics community required ongoing support and coordination among genetic counselors in Las Vegas and Reno. Regular communication was essential to track cases, ensure proper referrals to pediatric geneticists, and, when possible, conduct genetic testing during pregnancy. Establishing prenatal, cardio-genetics, neurogenetics, and congenital hearing loss case conferences would facilitate collaboration among stakeholders across Nevada. These efforts required sponsorship and a dedicated coordinator to maintain organization and ensure effective communication within the genetics community.

Chair Logan-Parker thanked Dr. Mark Nunes for his participation.

4) <u>FOR POSSIBLE ACTION:</u> Discussion and possible action in response to Senate Bill (SB) 78 of the 83rd Legislative Session, that revises provisions relating to boards, commissions, councils and similar bodies. – *Council Members*

Chair Logan-Parker discussed Senate Bill 78 and the potential elimination of the Nevada Rare Disease Advisory Council (RDAC). The bill proposed revisions to Nevada's boards, commissions, and advisory councils, raising concerns due to the RDAC's critical role in representing the rare disease community, which often faced challenges such as limited access to care and delayed diagnoses. The council's efforts had informed policymakers, with the upcoming release of the 2024 needs assessment data further highlighting its contributions. A request was made for the council to grant the chair authority to speak and testify on its behalf to ensure timely advocacy for policies supporting patients, caregivers, and

healthcare providers. Proactive engagement with lawmakers, stakeholders, and decision-makers was encouraged as new legislative issues emerged.

Council member Kim Anderson Mackey, serving as the Chairman for the Palliative Care and Quality of Life Council, acknowledged that the subcommittee discussed the broad scope of the proposed bill, which aims to eliminate inactive councils. The subcommittee indicated that they are not currently up for elimination, it may be moved under a different branch.

Council member Dr. Ihsan Azzam asked if the bill provided any benefits to eliminating the RDAC or merging it with another council.

Chair Logan-Parker explained that the bill aimed to streamline administrative burdens and processes, particularly for professional licensing boards such as the Board of Nursing and the Board of Medical Examiners, which had faced scrutiny for delays in licensure. Another part of the bill focused on volunteer organizations like the Nevada Rare Disease Advisory Council, which, despite being run by unpaid volunteers, required administrative support and resources. Recommendations from the Cure 4 The Kids Foundation suggested separating volunteer organizations from licensing boards to prevent them from being grouped with fee-based entities. Although the council initially considered dissolving due to inefficiencies, it was acknowledged that significant progress had been made, leading to a more successful and effective council. The National Organization for Rare Disorders (NORD) recognized the Nevada Rare Disease Advisory Council as the most efficient and effective in the country and expressed willingness to testify in support of its exclusion from elimination under SB 78.

**Council member Kim Anderson Mackey propose a motion that Chair Annette Logan-Parker would be granted the authority to formally speak for the Nevada Rare Disease Advisory Council and testify on behalf of the Nevada RDAC on legislative matters that impact the Council and the rare disease community during this 83rd legislative session in the state. Council member Valerie Porter seconded the motion to approve. There were no objections. A quorum voted to approve Chair Annette Logan-Parker would be granted the authority to formally speak for the Nevada Rare Disease Advisory Council and testify on behalf of the Nevada RDAC on legislative matters that impact the Council and the rare disease community during this 83rd legislative session in the state**

5) INFORMATIONAL ITEM: Review and discussion of the 'While You Wait' Needs Assessment Report on Year One. – *Chair Annette Logan Parker*

Chair Logan-Parker announced key findings from the needs assessment report, conducted from February 28 to December 31, 2024, which gathered 206 complete survey responses, with 72 participants not completing the survey. The survey highlighted challenges faced by Nevada's rare disease community, including barriers to healthcare access, financial support, insurance navigation, and education. Most respondents were parents of children with rare diseases, reflecting the critical role of caregivers across diverse age, race, ethnicity, and income groups. The report identified 151 unique rare disease diagnoses, emphasizing the diversity and complexity of rare diseases in Nevada. Significant delays in diagnosis and treatment were common, with many patients consulting five to seven doctors before receiving a

confirmed diagnosis, leading to prolonged wait times, worsened symptoms, and increased emotional and financial strain. Rural families faced additional challenges, including long travel distances for specialized care, while many families reported insurance denials for essential services and annual healthcare costs exceeding $5,000, with some surpassing $20,000. Out-of-pocket expenses for medical travel, genetic testing, and specialized therapies created significant financial stress. Children with rare diseases often struggled to obtain necessary educational accommodations, while young adults experienced disruptions in care during the transition from pediatric to adult healthcare due to gaps in provider knowledge and planning. Limited access to research and clinical trials was also evident, with over 60% of respondents unaware of available opportunities, further hindered by geographic distance, restrictive eligibility criteria, and insufficient provider awareness, especially in rural areas. The report included examples of families expressing frustration over delayed diagnoses and the need to fundraise due to denied treatments, underscoring the urgency of addressing barriers faced by the rare disease community at the state level.

7) INFORMATIONAL ITEM: Update on the approved letters of support for BDRs 123, 124, 38-218, 57-344, and 40-343 of the 83rd Legislative Session (2025) that will be submitted to the Nevada State Assembly – Health and Human Services and the Nevada Senate Health and Human Services. – *Chair Annette Logan Parker*

Chair Logan-Parker provided an update on the status of letters of support for BDRs 123, 124, 38-218, 57-344, and 40-343, with some already advanced to full Senate bills. The letters, reflecting the Council's position, incorporated data from the Nevada Rare Disease Advisory Council's needs assessment, emphasizing patient experiences and policy recommendations. Finalized letters were planned for submission to the appropriate legislative committees, with efforts made to monitor the progress of the bills, including hearings and testimonies. The Council's legislative engagement was highlighted as crucial for shaping policies that improve the lives of Nevadans with rare diseases, maintaining momentum and a strong reputation with stakeholders.

9) INFORMATIONAL ITEM: Council member information sharing announcements – *Council Members*

Chair Logan-Parker opened the floor for council member announcements.

Kim Anderson Mackey discussed the Palliative Care and Quality of Life Council, established in 2017, which focused on educating professionals and the public about palliative care in Nevada. Over the past four years, the council had hosted four to five Continuing Education Unit/Continuing Medical Education (CEU/CME) events annually, with 200 attendees each year and 50% new participants. A planned visit to Carson City on April 16th aimed to provide information during National Advance Care Planning Day, coinciding with the council's spring remote CEU/CME event. The council also reviewed AB 161 from the 83rd legislative session, a hospice regulation bill introduced by Dr. Edgeworth, addressing the growth from fewer than 40 licensed hospices in 2005 to 292 in 2024. The subcommittee was evaluating the bill, with expectations to support it while recommending adjustments before presenting it to the full council.

Chair Logan-Parker asked how the council managed its CMEs.

Kim Anderson Mackey answered that Project ECHO at UNR was consistently partnered with to provide CEUs/CMEs to social workers, nurses, physicians, and other professionals due to its convenience and extensive offerings. The program offered numerous free CEUs across various disciplines statewide, supported by a dedicated team, making it an ideal model for the Rare Disease Council to consider for annual education initiatives.

10) PUBLIC COMMENT:

Chair Logan-Parker opened the floor for public comment.

Hearing none, Chair Logan-Parker moved on to adjourn the meeting.

11) ADJOURNMENT: – *Chair Logan-Parker*

Chair Logan Parker moved to adjourn and expressed appreciation for everyone on the council.

**Chair Logan-Parker moved to adjourn the meeting at 11:09 am.**