Rare Disease Advisory Council

State of Utah
Department of Health and Human Services
November 29, 2025

To: Health and Human Services Committee
From: Nathan Shepherd, Department of Health and Human Services
Subject: Bi-annual legislative report on the Rare Disease Advisory Council

Attached is the 2024-2025 report to the legislature of the Rare Disease Advisory Committee (RDAC) of the state of Utah.

It contains RDAC's purpose, background, executive summary and accomplishments. It details the actions of the following of RDAC's workgroups:

- Legislative
- Stakeholders and Communications
- Data and Challenges

The report concludes with recommendations for the State Legislature and the Department of Health and Human Services.

On behalf of Utah's RDAC, we submit the 2025 report to the legislature for review.

Purpose

In accordance with Utah Code 26B-1-402(4), the department shall report to the Health and Human Services Interim Committee about (1) the activities of the grantee and the council, and (2) recommendations and best practices regarding the ongoing needs of individuals in the state with a rare disease.

The statute requires a report to be submitted by November 30, 2021, and by November 30 of every odd-numbered year thereafter. This 2025 report responds to that requirement.

Background

During the 2020 session, the Utah Legislature passed HB106, establishing the Rare Disease Advisory Council (RDAC) adjunct to the Utah State Department of Health and Human Services (DHHS)[1]

The legislation tasked the RDAC with advising and providing recommendations to the state legislature and state agencies on improving care and services for people with rare diseases. The RDAC includes a representative from the DHHS, researchers and physicians specializing in rare diseases, rare disease patients and caregivers, and representatives from rare disease organizations.

The Orphan Drug Act defines a rare disease as a disease or condition that affects fewer than 200,000 individuals in the United States.[2]

Rare diseases are rare individually, but as a group they are more common than many people think. Approximately 7,000 rare diseases affect more than 30 million people in the U.S. An estimated 1 in 10 Utahns, or 350,000 people, are suffering from a rare disease.[3] Most rare diseases, whether it is an immune deficiency, a rare cancer, or a genetic disorder, do not have FDA-approved treatments.

For purposes of the RDAC, the term rare disease also includes patients with undiagnosed conditions. The challenges of these patients are unique and require special attention. Many who live with rare diseases face reduced quality of life due to the lack or loss of autonomy caused by chronic, progressive, degenerative, or life-threatening aspects of many rare diseases.

The RDAC is funded by a $9,500 appropriation. The RDAC statute includes a sunset date of July 1, 2026. The state legislature, however, during the 2025 interim session, repealed the sunset date,

[1] H.B.106 – 63rd Utah State Legislature (2020). Rare Disease Advisory Council. Retrieved from https://le.utah.gov/~2020/bills/static/HB0106.html
[2] H.R.5238 - 97th Congress (1981-1982): Orphan Drug Act. (1983, January 4). Retrieved from https://www.congress.gov/bill/97th-congress/house-bill/5238
[3] Genetic and Rare Diseases Information Center; National Center for Advancing Translational Sciences; FAQs About Rare Diseases: https://rarediseases.info.nih.gov/about. The population of Utah in 2022 was approximately 3.5 million.

thus reauthorizing the RDAC. BioUtah, Utah's trade association for the life sciences sector serves as the DHHS-appointed grantee to administer the RDAC. All RDAC members are unpaid volunteers.

Executive Summary

RDAC members share a vision and passion to better address the needs of rare disease patients and their caregivers and improve health outcomes.

The 2025 report details the accomplishments of the RDAC since the last report in 2023 and provides recommendations to the state legislature and state agencies. The RDAC has met eight times since 2023. To facilitate the work of the RDAC, subject-specific workgroups meet routinely between meetings of the full RDAC.

Some members initially appointed to the RDAC have left the council. New members have been appointed to fill vacant positions and to build out the council's knowledge and skill base.

A major focus of the RDAC has been the development and launch of the RDAC website. The website, which is independent of the DHHS, was over a year-long project which included site design, construction, and content decisions to ensure that the site provided information about the RDAC and its members, pertinent resources, and was user friendly. In addition, the RDAC embarked upon a larger communications strategy, including creation of a RDAC logo and social media profile. Plans are also underway to launch a RDAC newsletter and survey the Utah rare disease community to collect data needed to further support the RDAC's mission.

The RDAC Policy Guide has been updated again this year. The guide consists of a series of statements on policies critical to the rare disease community.

The guide's policy statements support the mission of the RDAC and provide a roadmap for taking steps that will help Utahns with rare diseases lead fuller, more productive and healthier lives. The guide is a dynamic document. Policies are modified or added as appropriate. In addition, the RDAC has been raising public awareness about rare diseases through speaking engagements, presentations and collaborations with stakeholders.

Consistent with the RDAC's independent advisory role, the RDAC is building a grassroots advocacy network, which as it grows, will help strengthen the voice of the council and Utah's rare disease community.

The RDAC was instrumental in organizing Utah's 2025 Utah Rare Disease Day and engaged Governor Spencer Cox in observing the day with a declaration.

The accomplishments and recommendations contained in this report will inform the focus and work of the RDAC in future years.

RDAC Accomplishments

Governance

1. The RDAC has held eight meetings since its 2023 report to the legislature.
2. Workgroups met routinely between full RDAC meetings to conduct business.
3. The RDAC combined two existing workgroups, Stakeholders and Partnerships, and Communications and Resources, into one workgroup titled the Stakeholders and Communications Workgroup.
4. The following five new members were added to the RDAC:
    - *Justine Case*, rare disease patient and advocate.
    - *Januel Gomez-Colon*, rare disease patient and quantitative analysis expert.
    - Cate Levy, genetic counselor at University of Utah's Huntsman Cancer Institute.
    - *Ava Szajnuk,* rare disease patient and student.
    - *Tanner Withers*, special needs planner.
    - *Nathan Shepherd*, Genetic Counselor, Utah Department of Health and Human Services.
5. The following members left their positions on the RDAC.
    - *Kristen Pauley*, genetic counselor at the Huntsman Cancer Institute.
    - *Melodie Weller*, PhD, assistant professor, School of Dentistry, University of Utah.
    - *Troy Evans,* rare undiagnosed patient.
    - *Heidi Maxfield*, member of the board of the Intermountain PKU & Allied Disorders, also National PKU Alliance.
    - *Kim Hart*, program manager, Newborn Screening, Utah Department of Health and Human Services
6. Changes were made to the Executive Committee as follows:
    - After four years of service as RDAC Chair, Lorenzo Botto, MD, stepped down from his position. He will serve as Immediate Past Chair. RDAC Vice Chair was unanimously voted to move up to Chair, while Sylvia Lam was unanimously voted in as Vice Chair. Gholson Lyon, MD, will remain Secretary until the end of 2025.

Legislative Workgroup

1. The Utah RDAC Policy Guide was updated this year. The annual update ensures that the RDAC is addressing timely patient interests, such as banning copay accumulators, providing robust newborn screening, and access to genetic testing and gene therapies.

2. A letter of introduction about the RDAC and a copy of the Policy Guide was sent to every legislature upon the opening of the 2025 legislative session. The purpose of the letter is to raise the visibility of the RDAC before lawmakers and share key policy priorities. (Addendum 2).

3. RDAC members attended Utah's General Sessions and Interim Sessions, focusing on hearings and bills relevant to its mission and that impact the rare and undiagnosed communities. Formal letters/statements were sent to committee chairs and committee

members to educate legislators about how certain bills would impact rare disease patients.

4. Highlights from 2024:
    a. The RDAC Legislative Workgroup tracked 31 bills, and sent a letter regarding Medicaid funding to the Health and Human Services Committee, urging that funding be maintained to protect the needs of rare disease patients.
    b. RDAC member Marian Furst testified about the importance of safeguards and protections for patients, and allowing pharmacists to substitute generics or biologics when necessary to ensure timely and cost-effective access to prescribed treatments.
    c. RDAC member Matt Pearl testified regarding the importance of funding Medicaid for rare disease patients and families.
    d. RDAC member Rebecca Yates was invited to present at an interim committee as a subject matter expert on copay accumulators.
5. Highlights from 2025
    a. Tracked 31 bills, formal letters sent to committees regarding six bills (Examples in Addendum 3)
    b. RDAC members met with Utah Medicaid Director, Jennifer Strohecker. The discussion centered around resources available to the Utah rare disease community as well as the importance of newborn screening, genetic testing, gene therapies, and stable Medicaid funding for rare disease patients. A continued dialogue is anticipated.
    c. RDAC member Justine Case advocated for preserving the integrity of Utah's Newborn Screening Program, met with legislators, testified at HHS hearing.
    d. RDAC members Januel Gomez and Gina Szajnuk educated legislators about the importance of Medicaid waivers for supporting the independence of disabled rare patients and their ability to work, which provides career growth options.
    e. RDAC members attended public meetings, and collaborated with other advocacy organizations, including but not limited to: Utah All Copays Count Coalition, Medicaid Advisory Council, Newborn Screening Advisory Council, and the Legislative Coalition for People with Disabilities, as well as other patient and medically focused organizations in Utah. These engagements help to raise public awareness about rare disease and provide a foundation for collective advocacy.

Stakeholders and Communications Workgroup

Stakeholders

1. RDAC outreach resulted in the following guest speakers at RDAC meetings:
    a. Jennifer Strohecker, PharmM (Utah Medicaid Director)
    b. Dr. Josh Bonkowsky, MD (Center for Personalized Medicine at Primary Children's Hospital/Gene Kids)
    c. Tracy Meier, National Ability Center
    d. Annie Kennedy and Dylan Simon, Every Life Foundation
    e. National Organization of Rare Disorders (NORD) representatives
2. RDAC members attended the RDAC National Leadership Meetings NORD Conference to learn and share best practices.
    a. 2023: Dave Viskochil and Marian Furst
    b. 2024: Lorenzo Botto and Sylvia Lam
    c. 2025: Matthew Pearl and Alex Pearl
3. Community Outreach, Organizing Rare Disease Day Events
    a. The RDAC organized and implemented the 2025 Utah Rare Disease Day Event
        i. Requested and received Governor Spencer Cox's formal declaration for 2025 Utah Rare Disease Day. View the declaration HERE.
        i. Rare Disease Day event, "An Evening of Hope", held on at Recursion, in Salt Lake City. Watch excerpts of the 2025 Utah Rare Disease Day event HERE.
        ii. Speakers included the following:
        Kelvyn Cullimore, President and CEO, BioUtah
        Sylvia Lam, Utah RDAC
        Chris Gibson, Recursion
        Aimee Edwards, Executive Director, BioHive
        Tracy George, Chief Scientific Officer, ARUP Laboratories
        Ava Szajnuk, Undiagnosed Rare Disease Patient, Rare and Undiagnosed Network (RUN)
        Josh Bonkowsky, MD PhD, Chief, Division of Pediatric Neurology & Director, Primary Children's Center for Personalized Medicine, University of Utah
        Rachel Palmquist, University of Utah Health, Intermountain Genetic Counselor, Primary Children's Gene Kids
        Michael George, Undiagnosed Rare Disease Patient, Rare and Undiagnosed Network (RUN)
        Stacey L. Clardy, MD PhD FAAN, MD PhD, Associate Professor of Neurology, University of Utah & Salt Lake City VAMC
        Najat Khan, PhD, Recursion Chief R&D Officer and Chief Commercial Officer

4. RDAC vice chair, Gina Szajnuk, presented on behalf of the RDAC before the Utah Life Sciences Innovation Caucus on the first day of the legislative session, January 17, 2025.

5. RDAC members attended local community and professional events to promote awareness about rare and undiagnosed disease, and forge partnerships. Examples include:
    a. 2024 Utah Life Sciences Summit, Nov. 2024
    a. 2025 Utah Life Sciences Summit, Nov. 2025
    a. 2025 Wilson Sonsini, Entrepreneur & Investment Life Sciences Summit, March 2025
    b. 63rd Western Intermountain Neurology Organization (WINO) Conference, April 2025
    c. BioHive Cares: Support Youth in STEM! May 2025
    d. United Angels Foundation, Walk with Angels, May 2025
    e. Uniting Women's Voices, Women in Technology and Science (WITS), Recursion Pharmaceuticals, June 2025
4. ARUP Laboratories, Utah Patient & Partners Coalition, June, 2025
4. Canyon Rim Cares Day of Service, June, 2025
5. The Sumaira Foundation's Rare Neuroimmune Patient Day, University of Utah, August 2025

Communications

1. A task force of the Stakeholders and Communications Workgroup led the RDAC in reviewing RDAC logo options. A decision on a final logo was approved by the full RDAC.
2. The Utah RDAC website was launched. The website includes RDAC member bios, links to local and national resources for Utah rare disease patients, the Policy Guide, and a listing of events, such as Utah Rare Disease Day and RDAC meeting dates. In addition, the website allows for individuals to engage with the RDAC. The website is a significant deliverable for the RDAC this reporting period. The DHHS indicated that the agency would no longer host the website given the independent function of the RDAC. A task force consisting of a subset of RDAC members worked with a website developer to design and deploy content. Work to enhance the website is ongoing.

3. A social media presence has been established on LinkedIn, Facebook, and Instagram. A quarterly newsletter as well as video content is contemplated. The overall goal of the workgroup is to increase visibility, credibility, increase awareness, and seek partnerships.

4. The Utah RDAC joined RDAC representatives from New Hampshire and Minnesota as part of a 90-minute workshop presented at the 2025 American College of Medical Genetics Annual Meeting entitled; “Rare Disease Advisory Councils – State Advocacy for the Rare Disease Community.” A representative from the National Organization for Rare Diseases also served on the panel. Dr. David Viskochil of the URDAC served as the

moderator and Dr. Lorenzo Botto delivered a powerpoint presentation and served on a 4-member panel for discussion as part of the workshop presentation.

Data and Challenges Workgroup

1. **Current Barriers**
   a. The Data and Challenges Workgroup has been striving to collect data on the population and needs of individuals with rare diseases in Utah - both in scope and in lived experience - to inform policy, strengthen services, and improve healthcare outcomes. However, these tasks have faced significant obstacles, including participant turnover and data access limitations.
   b. Efforts to obtain primary data (as opposed to general estimates) on the number of individuals with rare diseases in Utah have been unsuccessful so far. Access to primary data has been financially and technologically unfeasible for a number of reasons, such as the fact that many rare diseases lack specific diagnostic codes in electronic medical records (EMR) and coding is not always uniform across EMR systems.

2. New Focus
   a. Considering these barriers, the workgroup is refocusing its efforts in two complementary directions.
   b. Identifying new ways to generate reliable estimates of rare disease prevalence in Utah.
   c. Adding a qualitative component to capture Utahns' lived highlight barriers to diagnosis, treatment, and ongoing care.

To advance this two-fold approach - combining data-driven estimates with firsthand community insight - the RDAC will finalize and pilot a self-report tool. The results of the pilot will help provide a more complete picture to date of rare disease in Utah and inform actionable recommendations.

Recommendations for state legislature

1. At least once a year, convene a meeting between the RDAC and the leadership of the House and Senate Health and Human Services Committees to discuss policies that affect individuals with rare diseases.
2. Outside the legislative session, convene an annual hearing with rare disease stakeholders, including the RDAC, to discuss ways to address challenges and barriers to care for the rare disease community.
3. Consider RDAC policy statements in shaping legislation. High priority should be given to the following crucial issues for the rare disease community:

   a. Scrutinize and reform pharmacy benefit managers (PBM) practices: limit abusive practices of pharmacy benefit managers by, at a minimum, increasing transparency requirements.

a. Improve access to genetic testing: facilitate access to diagnostic genetic testing under state-controlled health insurance policies. Early access to genetic testing allows the timely diagnosis of genetic rare diseases, which improves care and quality of life for affected individuals and caregivers and reduces long-term costs for payers.
b. Accelerate access to gene therapy: gene therapy is a medical treatment that offers cures or long-term improvements in health and quality of life to patients with serious or life-threatening rare diseases. Explore with stakeholders, including policymakers, state regulators, patients and industry, Medicaid value-based payment arrangements or other reimbursement models that could advance access to and coverage of gene therapies.
c. Facilitate access to precision care: precision care is providing the right medication or agent to the right patient, at the right dose, at the right time, through the right route. Some patients with a rare disease may respond very favorably to a specific medication or agent, even though the medication is not yet approved for the rare disease.
d. Allow children to have the same treatment as adults for comparable diseases: children can develop the same rare diseases as adults. However, clinical trials have not or cannot be performed in children to establish FDA approval which can be a necessary requirement for health insurance to cover the cost. Children should be provided coverage for therapy if care providers treat and monitor the effects of therapy.
b. Reinforce or improve support for Medicaid: many rare disease patients are state Medicaid beneficiaries. Individuals diagnosed with a rare disease at a young age are likely to require medical care and incur greater medical expenses over a lifetime. Restructuring Medicaid benefits and eligibility in ways that restrict services could cause harm to people with rare diseases and increase overall costs to the healthcare system.
e. Consider rare diseases in public health/COVID-19 policies: consider the needs of the Utah rare disease community when implementing legislation related to public health, public health emergencies, and participation of rare disease patients in society.

Recommendations for DHHS

1. Identify current DHHS programs and resources that support or could support the Utah rare disease community and examine barriers or gaps in services.
2. Together with the RDAC, develop a communication plan to widely disseminate current information on available resources for the rare disease community and their families.
3. Work with the RDAC to better understand the population with rare diseases, including the current prevalence and distribution of rare diseases in Utah. As part of the plan, identify the data needed to better inform public policy, current data gaps, and data systems available in Utah to address the data gaps.

Recommendations for insurance regulation and services

1. Meet with RDAC representatives to explore remedying barriers to coverage of testing and healthcare services for rare disease conditions.

Conclusion

In conclusion, the RDAC has produced a record of accomplishments and identified on-going challenges. In addition, the RDAC has developed recommendations for the state legislature and state agencies on critical issues, including the need for services, diagnostic tools, access to affordable treatments, funding, and identification of the population of individuals living with rare diseases in Utah.

The RDAC looks forward to working with the state legislature, state agencies and other stakeholders to implement the objectives of this report and help drive positive change for Utahns with rare diseases.

Addendum 1
Council Membership

Addendum 2
Policy Guide and Letter of Introduction to the Legislature

Addendum 3
Examples of letters to the State Legislature 2025

Addendum 1
Council Membership

Note: Italicized text reflects statutory criteria for RDAC members.

Executive Committee
Two representatives from one or more rare disease patient organizations that operate in the state:
RDAC Chair, Gina Szajnuk
A rare disease advocate and care giver to her children with rare disease. Co-founder and executive director of the Rare and Undiagnosed Network (RUN) organization, a 501(3)C non-profit.

RDAC Vice-Chair, Sylvia Lam
Executive Director, United Angels Foundation (UAF), supporting parents and families of individuals with special needs diagnosis and provides education, resources and community support.

Researchers and physicians who specialize in rare diseases, including at least one representative from the University of Utah:
RDAC Secretary, Gholson Lyon, MD, PhD
Clinical Professor, Roseman University College of Medicine, Las Vegas, Nevada and South Jordan, Utah; and Professor (Adjunct), City University of New York. Board certified psychiatrist.

Members
Researchers and physicians who specialize in rare diseases, including at least one representative from the University of Utah:
Lorenzo D. Botto, MD (immediate past RDAC Chair)
Professor of Pediatrics in the Division of Medical Genetics, University of Utah and Attending Clinical Geneticist at the University of Utah and Primary Children's Hospital.

Pınar Bayrak-Toydemir, MD, PhD, FACMG
Medical Director, Molecular Genetics and Genomics and, Professor, University of Utah School of Medicine and ARUP. American Board of Medical Genetics and Genomics.

Ron Day, MD
Retired physician with extensive experience in rare diseases; President of Blow by Oxygen Foundation which advances ideas for oxygen delivery to children with pulmonary hypertension.

Dave Viskochil, MD, PhD
Professor of Pediatrics in the Division of Medical Genetics, University of Utah and Attending Clinical Geneticist at the University of Utah and Primary Children's Hospital. Currently, chair of the Utah Newborn Screening Advisory Committee.
Sally Jo Zuspan, RN, MSN
Director of Research for the Data Coordinating Center at the University of Utah supporting multiple research networks, including oversight for multi-institutional clinical trials.

Two individuals who have a rare disease or are the parent or caregiver of and individual with a rare disease:
Justine Case
Rare disease patient and patient advocate with expertise in healthcare administration. Pursuing a master's degree in social work.

Kira Chaney
Rare disease patient with a bachelor of science in Anthropology from the University of Utah's College of Social and Behavioral Sciences and a minor in integrative human biology.

Januel Gomez-Colon
Rare disease patient and University of Utah graduate in Quantitative Analysis of Markets & Organizations, with an emphasis in Finance.

Steven Eliason
State legislator and parent of a rare disease child.

Marian Furst
Rare disease patient and active member of the Immune Deficiency Foundation. Retired patent and trademark attorney, with experience in patent preparation, prosecution and negotiating intellectual property licenses.

Cate Levy
Genetic counselor at Huntsman Cancer Institute, University off Utah.

Matt Pearl
Rare disease patient with Faconci Anemia (FA) and leader in the FA community. Served as a NORD ambassador.

Ava Szajnuk
Rare disease patient and student. .

Tanner Withers
Special needs planner.

Rebecca Yates
Licensed insurance broker and owner of Ark Insurance Solutions, LLC. With a family history of Ehlers-Danlos syndromes, a rare inherited disease, Rebecca is both a patient and mother of rare disease children.

Representative from the Department
Nathan Shepherd, Genetic Counselor, Utah Department of Health and Human Services

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Grantee (BioUtah)
Kelvyn Cullimore
President and CEO
BioUtah

Denise Bell
VP, Programming and Government Affairs
BioUtah

Addendum 2

Policy Guide and Letter of Introduction to the Legislature

RDAC
Utah Rare Disease Advisory Council

January XX, 2025

Dear Senators and Representatives,

In 2020, the Utah legislature established the Rare Disease Advisory Council (RDAC) (Utah Code Annotated Section 26-1-41) to advise the state legislature and state agencies on improving access to treatment and services provided to individuals with a rare disease and identify best practices to improve the care and treatment of individuals in the state with a rare disease.

An estimated one in ten Utahns, or an estimated 350,000 women, men, and children in our state, are suffering from one of the thousands of rare diseases, from genetic conditions and immune deficiencies to cancers and more.

The RDAC is an independent advisory body housed under the Utah Department of Health and Human Services. Members include physicians, researchers, rare disease patients and caregivers, and representatives from rare disease organizations.

As part of the RDAC's charge to advise the legislature, we provide you with the attached policy guide, which covers the following topics:

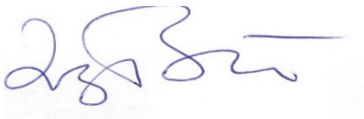
- Access to Genetic Testing
- Ban on Co-Pay Accumulator Programs
- COVID-19: Public Health Guidelines, Access to Vaccines and Treatments
- Gene Therapy
- Pharmacy Benefit Managers, and more.

A top priority for the RDAC in the 2025 session is the passage of legislation to ban copay accumulator programs. Copay accumulator programs significantly harm Utahns with rare diseases by preventing them from using manufacturer-provided copay assistance towards their deductibles and out-of-pocket costs, leading to drastically increased financial burdens and potentially forcing them to forgo necessary medications and care.

We are deeply grateful to the legislature for providing the rare disease community with this opportunity through the RDAC to help lawmakers and other stakeholders better understand and meet the challenges and needs of this constituency.

For more information, please contact the RDAC Chair, Dr. Lorenzo Botto, at lorenzo.botto@hsc.utah.edu, or the RDAC Vice Chair, Gina Szajnuk, at ginaszajnuk@gmail.com.

Sincerely,



Lorenzo D. Botto, MD, FACMG
Chair, RDAC
Professor of Pediatrics
Director, Penelope Undiagnosed and Rare Disease Program
PI, Intermountain West Clinical Site of the Undiagnosed Disease Network
Division of Medical Genetics, Department of Pediatrics, University of Utah

Gina Szajnuk
Vice Chair, RDAC
Co-founder and Executive Director, Rare and Undiagnosed Network (RUN)

RDAC
Utah Rare Disease Advisory Council

Improving the Lives

OF RARE DISEASE PATIENTS

POLICY GUIDE

Above: Utah siblings Matt and Alexandra Pearl both have Fanconi Anemia, a very rare genetic condition.

RDAC
Utah Rare Disease Advisory Council

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INTRODUCTION

Welcome to the 2023 Utah Rare Disease Advisory Council (RDAC) Policy Guide. The Guide is a compilation of policy statements about issues of critical importance to Utah's rare disease patients, their families and caregivers. Those who live with rare diseases may face reduced quality of life due to the burdens of their diseases as well as a lack of autonomy caused by chronic, progressive, degenerative, and/or life-threatening aspects of many rare diseases. Patients, as well as their families and caregivers may face burdens, including a lack of financial and other resources, along with disability resulting from the diseases and their treatments.

The RDAC was established by the Utah State Legislature with a mission to provide an in-depth understanding about rare disease for government officials and policymakers. The goal is to address barriers to resources, proper treatment and care.

The Guide's policy statements support the mission of the RDAC and provide a roadmap for taking steps that will help Utahns with rare diseases lead full and healthy lives. The Guide is a dynamic document. Policies will be modified or added as appropriate.

ABOUT THE RARE DISEASE COMMUNITY

Rare diseases are rare individually, but as a group they are more common than many people think. Approximately 7,000 rare diseases affect more than 30 million people in the U.S. An estimated one in ten Utahns, or an estimated 350,000 women, men and children are suffering from a rare disease.

Whether it is an immune deficiency, a rare cancer, or a genetic disorder, medical care for a rare disease is chronic and costly. Moreover, although the FDA has approved hundreds of drugs for rare diseases, most conditions do not have FDA-approved treatments. Drug, biologic and device development in rare diseases is challenging for many reasons, including the complex biology, the lack of understanding of many rare disorders and the absence of financial motivation due to small market sizes.

ABOUT THE RDAC

The RDAC was formed under HB 106 during the 2020 legislative session of the Utah Legislature (Utah Code Annotated Section 26-1-41) "to advise the state Legislature and state agencies on improving access to treatment and services provided to individuals with a rare disease; make recommendations to the Legislature and state agencies on improving access to treatment and services provided to individuals with a rare disease; and identify best practices to improve the care and treatment of individuals in the state with a rare disease."

The RDAC includes a representative from the Utah Department of Health and Human Services, researchers and physicians who specialize in rare diseases, rare disease patients and caregivers, and representatives from rare disease organizations.

For more information, please contact Lorenzo Botto, M.D., RDAC Chair, at lorenzo.botto@hsc.utah.edu. or Gina Szajnuk, RDAC Vice Chair, at ginaszajnuk@gmail.com

Approximately 7,000 rare diseases affect more than 30 million people in the U.S.

BAN ON COPAY ACCUMULATOR PROGRAMS

RDAC POSITION

Copay accumulator programs impose a considerable burden on the community of people with rare diseases, adversely impacting their health and well-being. In addition to increasing out-of- pocket costs, the resulting skipped treatments can cause adverse health outcomes for patients, including rare disease patients. These adverse health outcomes may increase overall costs to other payers, such as state-sponsored health plans, insurers and pharmacy benefit managers. The RDAC recommends banning copay accumulator programs in the State of Utah.

ISSUE

For medical insurance coverage, many families and individuals have deductibles of thousands of dollars and may also have significant copays or coinsurance. Many patients, including patients with rare diseases, receive financial assistance from drug manufacturers and other third parties to help pay for their medications, which frequently include high-cost specialty medications that are used long-term. This financial assistance significantly helps these people meet out-of-pocket costs, including deductibles, copays and coinsurance, so patients can access proven interventions.

Some insurance companies have instituted copay accumulator programs that typically target specialty drugs for which third-party copay assistance is available. The accumulators prevent copay assistance from applying to a patient's out-of-pocket costs for health insurance deductible, copays and coinsurance. Consequently, patients who depend on third-party support must still pay the full copay, coinsurance, and/or deductible, despite a third-party having already paid it on their behalf. The result is unnecessary financial hardship and increased likelihood of treatment non-adherence, which can lead to a more advanced disease state and/or increased disability for patients.

Families and individuals, especially those who have rare diseases with high treatment costs, face decisions about whether to delay or postpone visits and care based on achieving their respective out-of-pocket costs. According to Doug Long, vice president of industry relations for IQVIA, prescription abandonment at pharmacies by patients has been fueled by cost shifting to patients, including copay accumulators. In 2021, 60% of prescriptions with patient monthly out-of-pocket costs of $125 or more were abandoned at pharmacies. Skipped medical visits, treatments and prescription pickups may result in deterioration of the patient's condition and lead to significantly increased costs for hospitalizations, office visits and treatments that otherwise would likely not have been incurred. In addition to increasing costs for additional care, deterioration of the patient's health may interfere with the ability of patients and caregivers to be contributing members of society.

The burden of copay accumulator programs is clear. Applying payments derived from all sources, including out-of-pocket and copay assistance program payments, to insurance deductible, copays and coinsurance would enable families and individuals to meet their financial obligations with minimal interruption of care.

COVID-19: PUBLIC HEALTH GUIDELINES, ACCESS TO VACCINES AND TREATMENTS

RDAC POSITION

There are many rare diseases that put Utahns at high-risk of severe disease from COVID-19. The lack of recognition of some rare diseases as a high-risk category for vaccination and treatment eligibility has had unintended, but serious, consequences. Delays in vaccination have impacted patient safety for many that were unable to get early vaccination. The RDAC asks that in future policy decisions and public messaging for COVID-19 (and for other pandemic preparedness and response), "rare diseases that may cause severe risks" be clearly considered a formal category of high-risk.

ISSUE

Throughout the COVID-19 pandemic, many members of Utah's rare disease communities have faced extreme disruption to their lives and medical care. For most rare diseases, there is no information that could inform risk of severe illness and complications from COVID-19 and patients have to rely on their healthcare provider's recommendations and on common sense to determine their risk. Utah public health messaging and policies regarding vaccination eligibility for those at high-risk has consistently been silent regarding the rare disease population, including those who may be at highest risk for severe disease and death.

The lack of recognition of rare disease as a high-risk category from vaccination and treatment eligibility has had unintended, but serious consequences.

For many, this lack of attention in messaging and eligibility requirements has resulted in confusion, hesitancy and significant delays in getting vaccinated. Because it wasn't clear that rare disease patients were identified as high-risk, this population faced a quandary - risk getting turned away at vaccination sites or delay vaccination and potentially endanger their health.

Subsequently, many of these patients remained unvaccinated and unprotected despite a physician recommendation that they receive a timely vaccine, even as other patients that were at less risk, but had a more common and specific condition were eligible.

The Utah State Department of Health and Human Services made recommendations based on the best available information at hand about how to quickly protect the patient populations at highest risk. However, while it may be not feasible to ask for familiarity of the approximately 7,000 rare diseases in the midst of a public health crisis, such conditions should be considered when decisions to prioritize patient groups are being made.

ACCESS TO GENE THERAPY

RDAC POSITION

Gene therapy holds tremendous potential to improve treatment and potentially cure many rare genetic diseases. The RDAC supports access to and coverage for gene therapy for Utah's rare disease community and our healthcare system at large. Engagement with key stakeholders—including patients, providers, payers, manufacturers, and policymakers—is vital to equipping Utah's healthcare system, and the many children and families it serves, to realize the tremendous potential of gene therapies for rare diseases.

The RDAC further encourages Utah's Medicaid Program to explore innovative payment approaches, such as value-based payment arrangements, that could facilitate availability and utilization of life-changing treatments and, in some cases, cures for those with rare diseases.

ISSUE

Gene therapy is a new generation of medicine where a functioning gene is delivered to a patient to treat and potentially cure a genetic disease. Through gene therapy, the underlying cause of a genetic disease can be targeted and corrected, potentially with a one-time treatment.

1 in 10 Utahns, or an estimated 350,000 women, men and children in the state, are afflicted by a rare disease.

Gene therapy holds tremendous promise for the **estimated 1 in 10 Utahns, or an estimated 350,000 women, men and children in the state, who are afflicted by a rare disease**[1]. If you don't yourself have a rare disease, you likely know a family member or friend with a rare disease—whether it is an immune deficiency, a rare cancer, or a genetic disorder. Rare diseases are often debilitating, life-threatening and lack alternative treatment options.

Unlike traditional medicines or therapies, which require frequent administration and focus on managing symptoms, gene therapy is designed as a potential one-time treatment aimed at targeting the underlying cause of a disease at the cellular level.

Gene therapy could deliver transformational improvement in health and quality of life for Utahns and their families and caregivers, as well as provide substantial value for Utah's healthcare system and communities.

- **Utahns Living with Rare Diseases**: Eliminate or reduce clinical burden of disease, improve quality of life and increase life expectancy.
- **Utah's Families and Caregivers**: Alleviate the emotional and physical burden of care, enabling work and socialization and facilitating family relationships.
- **Utah's Healthcare System**: Reduce the need for chronic treatment, lower occurrence of disease-related complications and decrease healthcare utilization.
- **Utah's Communities**: Contribute to

1 For more information about PBM reforms implemented in other states, see: https://insidesources.com/rein-in-profiteering-by-drug-industry-middlemen/ article by David Balto, April 11, 2022. https://nhjournal.com/pezzillo-the-real-cause-of-the-high-price-of-prescription-drugs-pbms/ article by Rich Pezzillo, March 24, 2022. https://www.pbmaccountability.org/ accessed on September 6, 2022.

longer, healthier and more productive lives, with increased contributions to schools, workplaces, our economy and other aspects of Utah's local communities.

- **Utah's Economy**: Reduce direct and indirect costs of rare disease to Utah's economy, including by decreasing chronic care costs and disability support, while increasing economic contributions from rare disease patients who are freed from symptoms. **It's estimated that the total economic burden of rare disease in the U.S. is over $966 billion**[2].

While Utah may only comprise a portion of that economic burden, **each year an estimated 150 Utahns born are diagnosed with a rare disease**[3], resulting in significant healthcare utilization and economic impact. Gene therapies could benefit Utah's healthcare system in transformative and sustainable ways, offering potential to avoid years—and potentially lifetimes—of burden associated with chronic disease management for severely debilitating and life-threatening diseases.

To illustrate, consider **Duchenne muscular dystrophy** (DMD) and **hemophilia**. In the U.S., the total annual burden of illness per DMD patient is approximately $121,000[4]. Twenty-seven percent of caregivers said they reduced working hours or stopped working completely to care for a relative with DMD[5]. For hemophilia, the overall lifetime cost of treating moderately severe to severe hemophilia ranges from $19 million to $23 million per patient[6]. Underemployment or part-time employment as a result of American's living with hemophilia is estimated to cause almost $4 million in societal losses per year[7].

Gene therapies that treat the underlying cause of DMD, hemophilia and other rare diseases could help Utah avoid these significant costs. Importantly, despite the upfront costs of gene therapies, **projections indicate that they will not threaten health system financial sustainability**. Despite an expected increase in approved therapies, projected 2030 gene therapy spending in the U.S. is only **0.2% of total healthcare spending**[8].

SUPPORTING ACCESS TO GENE THERAPY

There are approximately **7,000 known rare diseases and 95% of them have limited or no treatment options. Fifty percent of people affected by rare diseases are children. With more than 500 gene therapies in clinical development**, all Utahns could benefit from improved access to these potentially life-saving cures and treatments for their children and families.

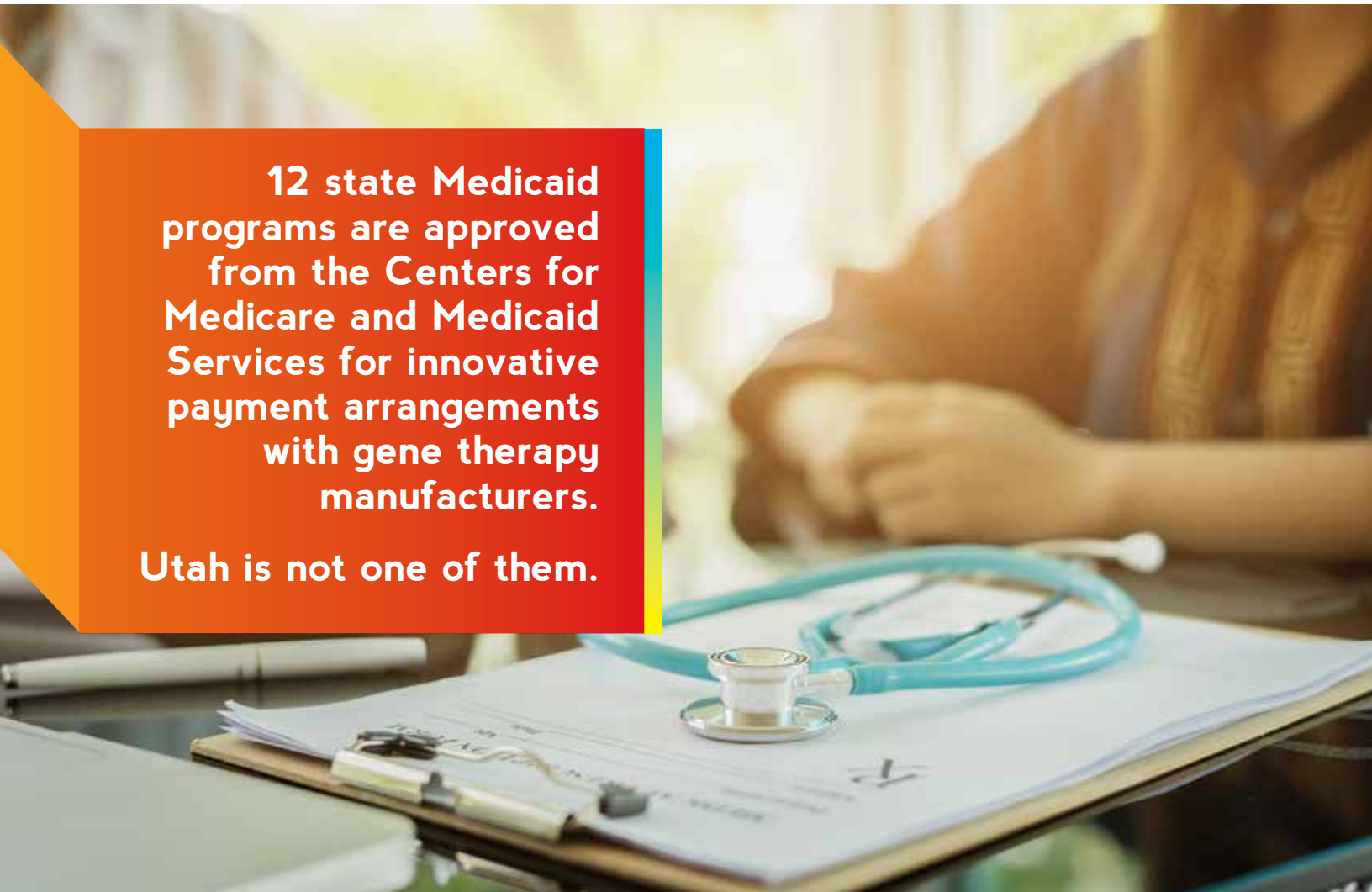
Because Medicaid and Medicare cover a significant share of those with disability status, these payers are expected to be the prominent payers for gene therapies. As more and more innovative gene therapies are discovered for rare diseases, Utah's Medicaid Program, along with Medicare, has

2 EveryLife Foundation. The National Burden of Rare Disease Study. February 25, 2021. https://everylifefoundation.org/burden-study/.
3 EveryLife Foundation for Rare Diseases. Newborn Screening – Utah. https://everylifefoundation.org/states/utah/.
4 Landfeldt, Erik, et al. The burden of Duchenne muscular dystrophy: an international, cross-sectional study. Neurology 2014;83(6):529-536.
5 Ibid.
6 Rind et al. Valoctocogene Roxaparvovec and Emicizumab for Hemophilia A: Effectiveness and Value; Evidence Report. Institute for Clinical and Economic Review, October 16, 2020. https://icer.org/wp-content/uploads/2020/10/ICER_Hemophilia-A_Evidence-Report_102620.pdf. Accessed May 2022. Li et al. Adult lifetime cost of hemophilia B management in the US: payer and societal perspectives from a decision analytic model. J Med Econ. 2021;24(1):363-72.
7 Chen, S. Economic Costs of Hemophilia and the Impact of Prophylactic Treatment on Patient Management. American Journal of Managed Care. Am J Manage Care. 2016;22:S126-S133.
8 Young, M., et all. Durable cell and gene therapy potential patient and financial impact: US projections of product approvals, patients treated, and product revenues. https://doi.org/10.1016/j.drudis.2021.09.001.

an opportunity to develop innovative policy to ensure children with debilitating and life-threatening conditions can access these potentially transformative therapies.

One barrier for State Medicaid Programs, in particular, is the actuarial risk of paying for gene therapy treatments. Compared to traditional chronic therapies, gene therapy treatment comes with large upfront costs, which are counterbalanced by significant, potentially life-long reductions in healthcare utilization and increases in productivity. Anticipating the potential budget impact of such new therapies on Medicaid programs is therefore of utmost importance. Consideration should be given to the need for sufficient annual appropriations and the use of innovative payment approaches, such as annuity or pay for performance contracts with gene therapy manufacturers.

Stakeholders from across the healthcare continuum are exploring alternative, innovative approaches to paying for these upfront costs while realizing the long-term benefits of treatment with gene therapies. Currently, 12 state Medicaid programs have sought and gained approval from the Centers for Medicare and Medicaid Services to enter into innovative payment arrangements with gene therapy manufacturers. Utah is not one of them.



12 state Medicaid programs are approved from the Centers for Medicare and Medicaid Services for innovative payment arrangements with gene therapy manufacturers.

Utah is not one of them.

ACCESS TO GENETIC TESTING

RDAC POSITION

Early and accurate diagnosis through genetic testing is crucial to improving outcomes for children and adults with rare diseases. The RDAC supports expanded access, funding and healthcare coverage of genetic testing for all Utah patients that could benefit from it.

ISSUE

Rare diseases represent an underserved and undercounted public health crisis. Most rare diseases have a genetic basis, and genetic testing, such as exome sequencing, whole genome sequencing and disorder-specific genetic panels, play an increasingly important role in the diagnosis and treatment of rare diseases.

Despite the importance of genetic diagnostics, there are still significant barriers for many rare disease patients that limit or prevent access to appropriate testing. The average length of time from the onset of recognizable symptoms to diagnosis of a rare disease in the U.S. is eight years. Misdiagnosis and delays in diagnosis carry high costs, including financial costs in terms of healthcare spending, and the human costs of avoidable disease progression and disability. Early and accurate diagnosis through genetic testing is crucial to improving outcomes for those with rare diseases.

MEDICAID EXPANSION

RDAC POSITION

The RDAC supports reasonable efforts to expand Medicaid coverage in Utah, including expansion through Utah state plan amendments and Medicaid waivers from the Centers for Medicare and Medicaid Services.

ISSUE

One of the greatest concerns for rare disease patients and their caregivers is obtaining health insurance coverage that provides access to the health care system for diagnosis and treatment, and many rare disease patients rely on Medicaid for this access. Rare diseases create health care problems that are not faced by the general public, but patients and their caregivers also must deal with the more routine health care needs faced by the general population. Therefore, any expansion of Medicaid coverage that benefits Medicaid beneficiaries in general also benefits the rare disease community. Any expansion of Medicaid benefits that is specific for rare diseases also is obviously beneficial to the community.

MEDICAL NUTRITION AND NEWBORN SCREENING

RDAC POSITION

The RDAC supports access to medically necessary nutrition for all individuals diagnosed with phenylketonuria (PKU) and other genetic metabolic disorders diagnosed on the basis of newborn screening, as recommended by physicians, the National Institutes of Health, Utah Department of Health & Human Services, the American College of Medical Genetic and Genomics and Genetic Metabolic Dietitians International. In addition, the RDAC supports the requirement that every baby born in Utah hospitals should receive a routine newborn screening test around 24 hours after birth. Every individual diagnosed with PKU, or other Inborn Errors of Metabolism requiring medical nutrition should be provided the appropriate coverage without unnecessary denials and appeals, regardless of insurance status.

ISSUE

Phenylketonuria (PKU) is a rare inherited inborn error of metabolism in which the amino acid phenylalanine is not broken down in the body properly, resulting in harmful build up of phenylalanine in the blood. In the United States, PKU occurs in about 1 in 10,000-15,000 newborns. Untreated PKU results in severe neurological complications and irreversible brain damage, including permanent intellectual disability, seizures, delayed development, behavioral problems, and other psychiatric disorders. Most cases are detected shortly after birth by newborn screening, and treatment is started promptly. Treatment consists of a life-long, carefully controlled, phenylananine-restricted diet beginning during the first days or weeks of life. Newer FDA-approved medications may allow a few people with PKU to follow a diet that has a higher amount or unrestricted amount of protein.[1] [2] [3]

Recognition of the critical importance of newborn screening is imperative. Newborn screening saves lives. Early diagnosis by newborn screening, along with medical treatment of PKU, and other inborn errors of metabolism, is critical for maintaining the health of infants born with these conditions. When an infant is diagnosed with PKU or another inborn error of metabolism, it is essential to begin treatment immediately with a protocol that includes medically necessary food and formula to maintain good health. The need for treatment is lifelong, and with strict adherence to treatment, patients may be able to lead normal lives.

1 https://medlineplus.gov/genetics/condition/phenylketonuria/
2 https://rarediseases.org/rare-diseases/phenylketonuria/?filter=ovr-ds-resources
3 https://www.mayoclinic.org/diseases-conditions/phenylketonuria/symptoms-causes/syc 20376302#:~:text=Phenylketonuria%20(fen%2Dul%2Dkey,needed%20to%20break%20down%20phenylalanine

In the United States, Phenylketonuria PKU occurs in about 1 in 10,000-15,000 newborns.

The standard treatment for PKU is a low protein diet, severely restricting the offending amino acid, phenylalanine (phe). This diet requires collaboration with a medical team to regulate the person's protein intake and determine specific daily intake of medical foods for optimal nutrition and development. This is accomplished by consuming medical foods and formula modified to be low in protein for the provision of required calories.

The PKU diet is costly and unaffordable for most people. Utah has mandated insurance coverage for medical nutrition for the treatment of inborn metabolic errors (Utah Code Annotated Section 31A-22-623), yet 72% of insured Utah residents are on federally regulated plans that pre-empt state legislation on medical nutrition. Many Utah families do not have coverage for low protein formula or foods and thus cannot adequately follow treatment protocol or recommendations due to access issues.

In 2021, the Utah legislature funded the PKU Formula Program for children through an ongoing appropriation to the Department of Health. This restored a former program to cover formula for all Utah children with PKU through age 18. From 2009-2021, the program was restricted to children with PKU under the age of 6 due to budget restraints. However, the need for medical formula and medical foods is life-long and continues through adulthood. All Utahns who require medical nutrition for the treatment of inborn metabolic errors should have access to lifesaving medical nutrition regardless of insurance coverage.

ACCESS TO MEDICAL RECORDS

RDAC POSITION

The RDAC supports improvements to medical records systems to provide easier access to records from different facilities for patients and providers.

ISSUE

Many Utahns receive medical care from multiple providers in different hospital or health care systems. In general, each system has set up its own medical record database. Thus, it is not always straightforward for providers and patients to access patient records, and the formats of the records also vary and make it more difficult to compare information from various dates and facilities.

One frequently encountered situation is that provider notes, lab results, imaging results, and other records are not always easily shared between systems. As a result, tests may be repeated unnecessarily, provider and patient time may be used inefficiently, appropriate care may be delayed or skipped entirely, and inappropriate care may be delivered.

Rare disease patients tend to have complex medical histories and often see many health care providers, making the lack of consistent access to their records even more of a problem than for other patients and providers.

Facilitating information sharing between hospital and health care systems would increase efficiency in providing care and simplify the burdens that both providers and patients face in managing care. It would be preferable to have a single user interface, data presentation format, and login arrangement that providers and patients can use to access records ordered by different providers and obtained at different facilities. Of course, protections need to be included to guarantee patient privacy and compliance with HIPAA requirements.

PHARMACY BENEFIT MANAGERS

RDAC POSITION

Utah should implement Pharmacy Benefit Manager (PBM) reforms to decrease drug costs for patients, ensure access to local pharmacies, and improve pharmacy reimbursement practices. A multi-pronged approach is necessary to address:

- Higher drug costs driven by PBM rebate policies that don't pass discounts on to patients.
- PBM lack of transparency.
- PBM discrimination against non-network and independent pharmacies.
- Cost pressures that burden state-run insurance programs as a result of the PBM business model.

Individuals in the rare disease community tend to use multiple medications and relatively expensive medications to manage their health conditions. Thus, the abuses of PBMs weigh especially heavily on this population.

ISSUE

PBMs are middleman companies that interface with pharmaceutical manufacturers, health insurers, and pharmacies. Often, they are part of large conglomerates that own or are owned by insurers and/or pharmacy chains with vertically and horizontally integrated businesses. PBMs develop drug formularies with and for insurers. This process is informed by factors such as negotiated reimbursement rates with pharmacies and negotiated rebates with drug manufacturers. The process includes discounts and other fees paid by manufacturers as well as drug placement in formularies.

Rare disease individuals often rely on multiple and more expensive medications to manage their health conditions.

The largest PBMs are interconnected with other behind-the-scenes affiliates, such as pharmacy payment processors, group purchase organizations (rebate aggregators), specialty drug labelers (especially for biosimilars), and healthcare providers. These affiliates may be headquartered outside the U.S., removing transparency in their operations. Acquisitions and mergers during the last few years have resulted in three large PBMs controlling about 80% of the prescriptions filled in the U.S. Another three control an additional 10%.

The Federal Trade Commission issued a report in July, 2024, describing the detrimental effects of PBM structures and operations. The agency has since sued CVS Health's Caremark, Cigna's Express Scripts, and UnitedHealth's Optum Rx for anticompetitive supply chain practices, resulting in artificially inflated insulin prices for some patients. The U.S. House Oversight Committee also issued a report in July, 2024, about PBM practices increasing drug costs for payers and consumers.

DRUG FORMULARY DESIGN

Formularies may discourage or encourage the use of generic and biosimilar medications through placement in pricing tiers or excluding them from a formulary altogether. Drug formularies may be designed on the basis of contracts with drug manufacturers for rebates and often implement utilization management provisions such as prior authorizations, step therapy, and quantity limits. Utilization management may function in keeping drug costs lower, but it is being misused to create obstacles for timely patient access to medically necessary drugs.

SPECIALTY DRUGS

There is no uniform definition of "specialty drugs," which are usually handled by specialty

pharmacies. The designation may be based on price alone or on a need for special handling and administration. Specialty drugs account for 40-50% of pharmacy dispensing revenue, but about 2% of total Rx volume. PBMs sometimes classify drugs as "specialty" to limit patient access at pharmacies not owned by or associated with the PBM.

PATIENT COST SHARING

PBM formularies and/or the associated health insurance plans are set up to share drug costs with patients. The cost used to calculate the patient's share is generally the manufacturer's list price, which may be significantly higher than the price negotiated with the drug manufacturer (see examples in Appendix A of the House Oversight Report), and patients typically don't receive the benefit of the manufacturer's price concessions to PBMs. In addition, third-party patient assistance programs for some expensive medications are intended to lessen the cost burden for patients, but PBMs and insurers are increasingly using practices such as copay accumulators and copay maximizers to pocket the financial assistance payments without applying them toward patients' out of pocket expenses.

PBM RELATIONSHIPS WITH PHARMACIES

PBMs have contractual relationships with pharmacies in their networks and may have common ownership with those pharmacies. Pharmacies may be retail, where patients pick up their prescriptions; mail order, where prescription medications are shipped to the patient's designated address; or specialty, where drugs designated as specialty may be delivered to the patient or to a clinic or care provider.

Often, reimbursement rates for payments to pharmacies for dispensing drugs discriminate against out-of-network and independent pharmacies. Inflated reimbursement rates may be paid to affiliated pharmacies. The reimbursement rates may change frequently and be difficult for pharmacists to understand. Post-sale adjustments, or clawbacks, have been assessed to independent and out-of-network pharmacies, based on designated pharmacy performance criteria that are outside of the pharmacy's control, such as patient compliance evidenced by repeat prescription refills. Reimbursement rates may be independent of reasonable costs for pharmacies to acquire the drugs. PBMs may set maximum allowable costs (MACs) for pharmacy reimbursement, which may be less than what the pharmacy pays. These practices may have serious adverse effects on the ability of non-network and independent pharmacies to operate profitably and may decrease market stability. Further, pharmacies have little bargaining power in setting contractual terms.

PBMs may engage in practices that steer patient specialty drug purchases to specialty pharmacies controlled by the PBM. Pharmacy purchases of generic and biosimilar drugs may be reimbursed only if they are obtained through PBM-affiliated labelers, with exclusivity requirements that prohibit pharmacies from obtaining specialty drugs from other sources. For example, a PBM may implement "white bagging" and designate a required pharmacy for purchase of a drug, or it may implement "brown bagging" and require a patient to purchase a drug at a designated pharmacy and bring it to their health care provider for administration.

Brown bagging is prohibited in Utah under SB 193, enacted in 2023.

Ways PBMs are Being Bypassed Currently
A limited number of pharmacies are offering mail order availability for some generic medications, which provides lower-cost access without insurance. These pharmacies may be in network for a few insurance plans. Mark Cuban's Cost Plus offers selected medications at prices that include the pharmacy's acquisition cost for the medication, a 15% fee to Cost Plus, a pharmacy fee for filling the Rx, and the shipping cost. CivicaRx is a nonprofit company that works to reduce hospital drug shortages and make lower-cost generic and biosimilar outpatient medications available,

including by manufacturing medications.

Companies such as GoodRx and SingleCare offer coupons or discount cards to consumers for many medications at their participating pharmacies. RX Partner offers prescription discounts to PBMs and health insurance plans. RX Savings Solutions provides services to pharmacists and health plans to offer drugs at lower prices to consumers. These two programs appear to work outside of PBM/insurer formularies.

AbbVie, the manufacturer of Synthroid, name brand levothyroxine, has a program that provides medication by mail and outside of insurance coverage, for $25/month, considerably less expensive than what many insurers pass on to patients as copays or coinsurance, if their formularies even include Synthroid.

There are several problems with bypassing insurance in these ways for rare disease patients and others who take multiple medications and receive them from multiple pharmacies. These options are only available for the limited selection of drugs available from the entity that is offering alternative pricing. Although using these pharmacies can reduce costs significantly for patients, patient payments for drugs acquired outside of insurance coverage do not count toward the insurance out of pocket costs.

Dealing with multiple pharmacies can be complicated for patients and their caregivers. In addition, mail order purchase from pharmacies may not be appropriate when the medication is needed immediately, such as antibiotics. One often-provided pharmacy service when new prescriptions are filled is to check whether concurrent use of the new prescription is contraindicated with other medications the patient takes, either because one medication interferes with the efficacy of another, or because serious adverse events can occur with concurrent use. A pharmacy that provides only one medication to a patient may not have access to a list of other medications the patient uses, and a pharmacy that provides the patient's other medications likely doesn't have a record of the medication from the other supplier.

PBM REFORMS UTAH CAN IMPLEMENT

Utah has already implemented some measures to improve accountability and transparency of PBMs. The state can implement additional legislation and take other appropriate steps to address PBM-related issues listed below.

TRANSPARENCY OF PBM OPERATIONS

- Reporting requirements for total drug spend, manufacturer rebates, and other discounts received.
- Total pharmacy compensation (see SB 208, 2018).
- Point-of-sale pharmacy price concessions.
- Patient cost-sharing.
- Establishing maximum allowable cost list requirements.
- PBMs to report conflicts of interest.
- Manufacturer reporting.

PBM INCOME

- Prohibitions against spread pricing, where insurers are charged more than is reimbursed to pharmacies.
- Requirements for PBMs to pass rebates to the health plan.
- Requirements for PBMs to apply rebates at the point of sale or use rebates to lower insurance premiums to reduce enrollee costs.
- Increasing Medicaid control over pharmacy reimbursement rates.
- Prohibiting a PBM from reimbursing a pharmacy less than the national average drug acquisition cost (NADAC), a metric used by Medicare.
- Prohibiting a PBM from reimbursing an unaffiliated pharmacy less than an affiliated pharmacy.
- Setting standards for pharmacy audits and recoupments.

DRUG COST SHARING

- Copay caps, already implemented for insulin and considered for albuterol inhalers and epinephrine injectors.
- Requiring third party payments to be applied toward out-of-pocket costs (co-pay accumulator and copay maximizer bans).
- Controlling State Costs (Medicaid and other State-Run Programs)
- Utilization management, which can include lifestyle changes.
- Limitations on step therapy.

EXAMPLES OF RECENTLY PASSED LEGISLATION IN OTHER STATES

Idaho HB 96 (2024)
Strong focus on transparency and reporting, pass through pricing to consumers, prevents steering, pharmacy adequacy standards, defines specialty drug.

Massachusetts Senate No. 2520
Passed both Houses and in conference committee (7/25/24). The bill includes PBM licensing with the Department of Insurance, periodic auditing of PBMs by health insurers, bans spread pricing, bans point of sale and retroactive fees with assessment to PBM for violation, requires at least 80% of rebates to be passed on to consumers at point of sale, requires pharmacies to charge customers the lesser of applicable cost-sharing or pharmacy retail price, requires copay assistance to be included in enrollee cost sharing, imposes duty of good faith and fair dealing on PBMs when dealing with all parties they interact with in performing PBM services, requires PBMs to provide an adequate and accessible network for prescription drugs, requires PBMs to maintain a Maximum Allowable Cost list for generic prescription drugs and must reimburse an independent pharmacy for drugs at the same amount that the PBM reimburses PBM affiliates for providing the same pharmaceutical services, makes permanent the ability of consumers to use drug manufacturer coupons to pay for prescription drugs, procedure for notice and hearing if planned closure of a pharmacy will create pharmacy desert.

Pennsylvania PA HB 1993 (2024)
Amounts of rebates and payments received from drug manufacturers and how those payments were distributed by the PBM.

ABOUT THE RARE DISEASE COMMUNITY

Rare diseases are rare individually, but as a group they are more common than many people think. Approximately 7,000 rare diseases affect more than 30 million people in the U.S. An estimated one in ten Utahns, or an estimated 350,000 women, men and children are suffering from a rare disease.

Whether it is an immune deficiency, a rare cancer or a genetic disorder, medical care for a rare disease is chronic and costly. Moreover, although the FDA has approved hundreds of drugs for rare diseases, most conditions do not have FDA-approved treatments. Drug, biologic and device development in rare diseases is challenging for many reasons, including the complex biology, the lack of understanding of many rare disorders and the absence of financial motivation due to small market sizes.

ABOUT THE RDAC

The RDAC was formed under Utah Code Annotated Section 26-1-41, enacted during the 2020 Utah Legislative session to advise the state Legislature and state agencies on improving access to treatment and services provided to individuals with a rare disease; make recommendations to the Legislature and state agencies on improving access to treatment and services provided to individuals with a rare disease; and identify best practices to improve the care and treatment of individuals in the state with a rare disease.

The RDAC includes a representative from the Utah Department of Health and Human Services, researchers and physicians who specialize in rare diseases; rare disease patients and caregivers; and representatives from rare disease organizations.

For additional information, please contact Lorenzo Botto, M.D., RDAC Chair, at lorenzo.Botto@hsc.utah.edu.

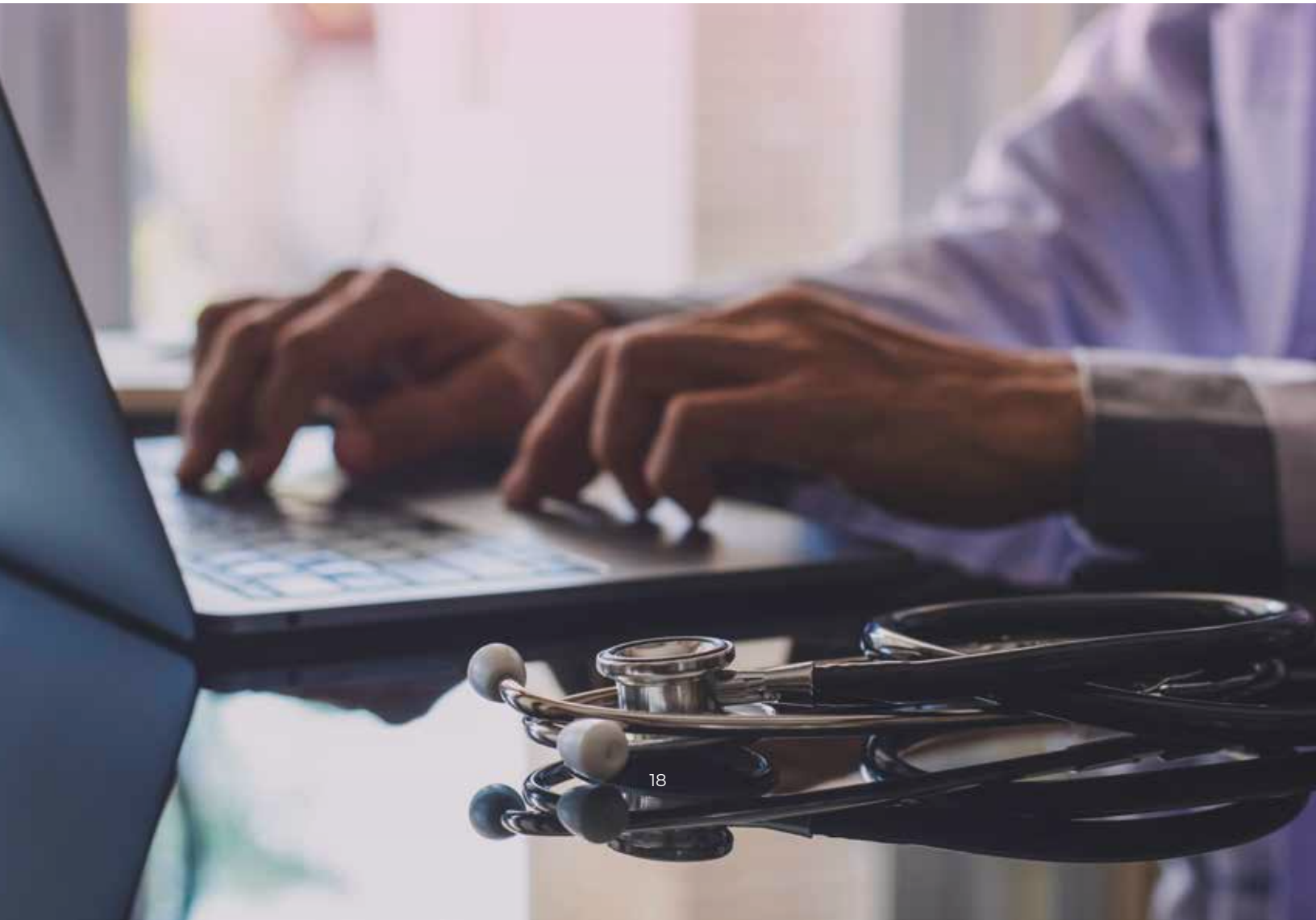
ACCESS TO TELEHEALTH SERVICES

RDAC POSITION

The RDAC supports the continuation of accessible telehealth services in Utah.

ISSUE

Telehealth services, provided in accordance with accepted standards of care and in appropriate situations, should continue to be available to all Utahns. Many rare disease patients have limited mobility, making it difficult for them to travel to medical appointments. Many rare disease patients are immunocompromised, which means they may face additional risks at medical facilities due to exposure to infectious diseases that they can't fight effectively. Thus, it is particularly important for rare disease patients to have continued access to telehealth services as appropriate.



VOTER BALLOT ACCESS

RDAC POSITION

The RDAC supports changes to the Election Code that would facilitate voting by people with disabilities, including changes that provide for alternative means of identity verification for people who cannot sign their names consistently due to a disability.

ISSUE

In Utah, most elections are conducted primarily by mail, with options available for in-person ballot submission and voting. The current system requires each voter to sign an affidavit that is included with the mail-in ballot, whether the ballot is returned by mail or dropped in a collection box. Poll workers compare the signature on the ballot to a previously obtained signature to verify the identity of the voter prior to counting each ballot. However, some disabled voters are unable to provide signatures or unable to provide reproducible signatures that a poll worker will find acceptable. Some rare disease patients are among these disabled voters. Disabilities that preclude use of signatures for ballot verification should not prevent the disabled voters from casting ballots.

It is also important to provide easy access to information about how people who are unable to provide consistent signatures can vote.

Perseverance

UTAH'S RARE DISEASE PATIENTS, INCLUDING THESE TEENS, DESERVE A CHANCE TO LIVE HEALTHY AND PRODUCTIVE LIVES.

RDAC
Utah Rare Disease Advisory Council

Utahns with rare diseases who participated in the Utah Rare and Undiagnosed Network teen campaign in 2016.

Addendum 3

Examples of letters to the State Legislature 2025

RDAC
Utah Rare Disease Advisory Council

November 19, 2024

Honorable Members of the Judiciary Interim Committee
Utah State Legislature
Utah State Capitol
350 State Street
Salt Lake City, UT 84103

Dear Members of the Judiciary Interim Committee,

The **Utah Rare Disease Advisory Council would like to voice its strong support for legislation, *Health Insurance Modifications*, scheduled for consideration in your committee that would impose a state ban on copay accumulator programs.** The legislation, sponsored by Representative Kera Birkeland, is on the committee's agenda for November 20.

The Utah RDAC was formed under HB 106 during the 2020 legislative session of the Utah Legislature (Utah code 26-1-41), to inform and advise the Department of Health and the Utah Legislature regarding issues confronting individuals with rare diseases. The council consists of rare disease patients, advocates, physicians, healthcare providers, and insurers.

The RDAC focuses mostly on challenges shared by many people across the spectrum of rare and undiagnosed diseases. One such challenge is that for these diseases, when treatable (and not all are), medical care is **chronic and costly**.

This is where the burden of copay accumulator programs become clear. Many families have deductibles of thousands of dollars and may also have significant copays or coinsurance. When financial assistance is provided to these families to help meet these deductibles and out of pocket costs so that individuals can access proven interventions, it is important that this payment assistance be allowed. However, under some insurance policies, such payments do not count towards the patient's deductibles. Instead, patients are still required to meet their deductibles or co-pays regardless of third party assistance.

The burden is significant – families, especially those with members who have rare diseases with high treatment costs, have to make decisions about whether to delay or postpone visits and care based on achieving their respective deductibles. **Applying copays derived from all sources, out of pocket copays and copay assistance programs, to insurance deductible would enable families to meet their financial obligations without interruption of care**.

Others have pointed out the unfairness of current copay accumulator programs. The Utah RDAC would like to emphasize how such programs, in addition to being unfair, also represent a significant burden to the Utah rare disease community. The *Health Insurance Modifications* legislation is an important step forward to provide some support to this community by ensuring that payment assistance for a patient's medication counts toward the deductible. For this reason, we join with many other patient and professional organizations in urging passage of the bill.

RDAC
Utah Rare Disease Advisory Council

February 24, 2025
To: Representative Bolinder
Cc: House HHS Committee
Re: H.B. 409 Medicaid Pharmacy Amendments

Dear Representative Bolinder,

The Utah Rare Disease Advisory Council (RDAC) was established by the Legislature in 2020 to advise legislators on policies affecting the rare disease community of Utah. https://utahrdac.org We are a collaboration of rare disease patients, advocates and medical professionals, and we wish to thank you and convey our support for HB409.

As many as 1 in 10 Utahns lives with a rare disease and frequently use Medicaid for pharmacy insurance coverage. It is our understanding the HB409 would reduce pharmaceutical costs to taxpayers by removing third party ACO, thereby streamlining pharmacy benefit distribution via a direct fee-for-service model. We support this increased efficiency and cost savings for all parties.

Thank you for your service and interest in patient needs and pharmacy benefit efficiency and affordability. Please feel free to contact any member of the RDAC if you have questions, or if we can be of service.

Sincerely,

Justine G. Case

Lead, RDAC Legislative Workgroup

Lorenzo D. Botto, MD, FACMG

Chair, RDAC
Professor of Pediatrics
Director, Penelope Undiagnosed and Rare Disease Program
PI, Intermountain West Clinical Site of the Undiagnosed Disease Network
Division of Medical Genetics, Department of Pediatrics, University of Utah

RDAC
Utah Rare Disease Advisory Council

February 26, 2025
To: Representative Dailey-Provost & Senator Escamilla
Re: H.B. 310 Disability Coverage Amendments

Dear Rep. Dailey-Provost & Sen. Escamilla,

The Utah Rare Disease Advisory Council (RDAC) was established by the Legislature in 2020 to advise legislators on policies affecting the rare disease community of Utah. https://utahrdac.org We are a collaboration of rare disease patients, advocates and medical professionals, and we wish to thank you and convey our support for HB310.

As many as 1 in 10 Utahns live with a rare disease, and many are disabled by their disease and rely on Medicaid. It is our understanding the HB310 would provide much needed services to support rare disease patients with disabilities to be able to work, increasing not only their quality of life and dignity, but also their financial capacity and independence without punitive withdrawal of essential supports.

Thank you for your service and interest in disability issues and rare patient needs. Please feel free to contact me, or any member of the RDAC if we can be of service.

Sincerely,

Justine G. Case
Lead, RDAC Legislative Workgroup

Gina Szajnuk
Vice Chair, RDAC
Co-founder and Executive Director, Rare and Undiagnosed Network (RUN)

Lorenzo D. Botto, MD, FACMG

Chair, RDAC
Professor of Pediatrics
Director, Penelope Undiagnosed and Rare Disease Program
PI, Intermountain West Clinical Site of the Undiagnosed Disease Network
Division of Medical Genetics, Department of Pediatrics, University of Utah

RDAC
Utah Rare Disease Advisory Council

February 26, 2025
To: Representative Daily-Provost & Senator Escamilla
Re: H.B. 310 Disability Coverage Amendments

Dear Rep. Daily-Provost & Sen. Escamilla,

The Utah Rare Disease Advisory Council (RDAC) was established by the Legislature in 2020 to advise legislators on policies affecting the rare disease community of Utah. https://utahrdac.org We are a collaboration of rare disease patients, advocates and medical professionals, and we wish to thank you and convey our support for HB310.

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Thank you for your service and interest in disability issues and rare patient needs. Please feel free to contact me, or any member of the RDAC if we can be of service.

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In closing, please know that rare diseases are rare individually, but as a group they are more common than many people think –**an estimated one in ten Utahns, or over 300,000 women, men, and children are suffering from a rare disease.** If you don't yourself have a rare disease, you likely know a family member or friend with a rare disease – whether it is an immune deficiency, a rare cancer, or a genetic disorder in a child.

Thank you for your consideration.

Sincerely,

The Executive Board, Utah RDAC

Lorenzo D. Botto, MD, FACMG
Professor of Pediatrics
Director, Penelope Undiagnosed and Rare Disease Program
PI, Intermountain West Clinical Site of the Undiagnosed Disease Network
Division of Medical Genetics, Department of Pediatrics
University of Utah

Gina Szajnuk
Co-Founder and Executive Director
Rare and Undiagnosed Network (RUN)

Gholson Lyon, MD, PhD
George A. Jervis Clinic and
Institute for Basic Research in Developmental Disabilities (IBR)

RDAC
Utah Rare Disease Advisory Council

March 3, 2025
To: Senator Johnson & Representative Hall
Re: S.B. 274 Health Insurance Preauthorization Revisions

Dear Sen. Johnson & Rep. Hall,

The Utah Rare Disease Advisory Council (RDAC) was established by the Legislature in 2020 to advise legislators on policies affecting the rare disease community of Utah. https://utahrdac.org We are a collaboration of rare disease patients, advocates and medical professionals, and we wish to thank you and convey our support for S.B. 274.

As many as 1 in 10 Utahns lives with a rare disease and frequently need medical procedures and treatments that require insurance preauthorization. It is our understanding the S.B. 274 would provide transparency and accountability for the cumbersome preauthorization process. We support your efforts to create this much needed monitoring.

Thank you for your service, interest in patient needs, and timely access to medical care. Please feel free to contact me (Justine Case) or any member of the RDAC if we can be of direct support.

Sincerely,

Justine G. Case
Chair, RDAC Legislative Committee

Lorenzo D. Botto, MD, FACMG
Executive Chair, RDAC
Professor of Pediatrics
Division of Medical Genetics, Department of Pediatrics, University of Utah

RDAC
Utah Rare Disease Advisory Council

May 27, 2025

To: Utah Dept. Health & Human Services, Division of Integrated Healthcare
Re: Proposed Section 1115 Demonstration Amendments, Community Engagement
Attn: Laura Belgique

The Utah Rare Disease Advisory Council (RDAC), established by the Legislature in 2020, advises the Legislature and State Agencies on providing services and care for individuals with rare diseases. We are a coalition of patients, advocates, community leaders, and medical professionals.

In this role, we wish to express our concerns and recommend revisions regarding the proposed Medicaid Community Engagement (CE) requirements. As many as 1 in 10 Utahns live with a rare disease, and we believe the current proposal could negatively affect this large and vulnerable population.

Disability Determination in Rare Disease. Many Utahns with rare or undiagnosed conditions rely on Medicaid and/or waiver programs. Diagnosing a rare disease often takes years and multiple specialist visits. While *undiagnosed*, the disease can be effectively invisible to administrative programs, due to its unclear nature and lack of appropriate coding. Still, these conditions are chronic and disabling. In this setting, establishing disability status can be difficult, especially with unknown or poorly understood diagnoses and fluctuating symptoms.

Due to these challenges, we are concerned that many individuals with rare or undiagnosed diseases may not meet the CE exemption criteria, resulting in the loss of essential Medicaid coverage.

• We urge you to consider the unique and significant challenges in establishing disability status in this population. Loss of coverage would jeopardize critical care and support.

Intermittent Disability and CE Requirements. Individuals with rare or undiagnosed diseases may be intermittently disabled or capable of limited work, yet unable to qualify for Social Security disability due to diagnostic uncertainty. Their conditions often involve variable or episodic symptoms.

• It is unclear how the CE program will address fluctuating disability status. We ask that such variability be explicitly recognized and accommodated

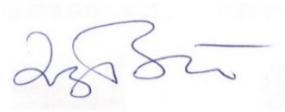
RDAC
Utah Rare Disease Advisory Council

Burden of Process and Risk of Coverage Loss. We are concerned about the administrative burden of the CE process and the risk of losing coverage due to procedural issues. The very act of proving exemption can become a barrier to care. According to Utah Medicaid Continuous Eligibility Unwinding data, 16,252 to 21,751 individuals *per month* were not renewed during the first six months due to procedural problems.

• We appreciate the inclusion of Member Support in the proposal. We recommend that this support include specific assistance for those navigating the exemption process to prevent unnecessary loss of coverage.

We thank you for your thoughtful consideration of the needs of Utah's rare and undiagnosed disease community. We offer our expertise as a resource and welcome continued dialogue toward collaborative solutions.

Sincerely, on behalf of the Utah RDAC,



Lorenzo Botto, MD
Executive Chair, Utah RDAC
Rare Disease Physician and Professor of Pediatrics
Division of Medical Genetics, University of Utah

Gina Szajnuk
Executive Vice Chair, Utah RDAC
Founder & CEO, Rare & Undiagnosed Disease Network
Undiagnosed Patient

Justine G. Case
Legislative Committee Chair, Utah RDAC
Rare Patient & Community Organizer