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Driven by science.
Aspire 4 Rare
Taking Policy Action on Rare Disease in the United States

Aspire4Rare: Prioritizing U.S. Policy Actions

Although great progress has been made since the enactment of the 1983 Orphan Drug Act, the landscape of rare disease treatment in the United States remains challenging. With more than 30 million Americans[1] navigating life with a rare disease, issues related to patient equity, access to treatments, and payment options are pressing.

The purpose of this report is to outline immediate, actionable, and realistic policy solutions supported by leading experts from across the rare disease spectrum, including clinicians, pharmacists, insurance providers, patient advocacy groups, and trade associations.

U.S. policymakers are encouraged to implement the policies outlined to ensure that millions of Americans receive the care and treatments they deserve.

Addressing systemic barriers in the rare disease community requires concerted effort and collaboration among all stakeholders involved in rare disease care. This is the foundational principle of **Aspire4Rare**, a global initiative which aims to provide an expert-developed framework for systematic change in the rare disease landscape, as outlined in the Aspire4Rare global framework **here**. This project was organized, funded and facilitated by UCB. UCB believes the rare disease community must work together to develop holistic, sustainable health systems for patients. Company representatives observed the discussion but did not influence the recommendations of the experts.

Overview: Top Recommendations to Address Rare Disease Policy Gaps

Patient Access to Specialists and Treatments

Solution I.
Alleviate the shortage of genetic specialists by addressing licensure requirements and telehealth service coverage.

Solution II.
Expand coverage decisions by addressing evidence gaps with strengthened post-market surveillance feedback loops.

Solution III.
Restore and enhance incentives to develop rare disease treatments.

Payment Options and Prior Authorization

Solution I.
Improve cost analysis approaches and implement flexible payment models.

Solution II.
Implement peer-to-peer review processes involving true medical peers.

Solution III.
Ensure patients have continuous access to rare disease treatments by reforming utilization management, step therapy, and prior authorization.

The recommendations presented in this report reflect the collective insights shared by the expert panel during their discussions. These recommendations are not to be interpreted as an endorsement of specific recommendations by individual panelists or the organizations they are affiliated with.

[1](NIH – National Institute of Health, 2024)

Patient Access to Specialists and Treatments

Rare disease patients often find themselves navigating a labyrinth of challenges to access the treatments and care they need. The majority – 95 percent[2] – of rare diseases lack approved therapies. Access issues are further exacerbated by geographic, demographic, and socioeconomic inequities. Patients in rural or underserved areas almost always face longer travel times to reach specialized care centers or providers, while those from disadvantaged socioeconomic backgrounds may struggle with the financial burden of treatment on top of burdens like the cost of food, housing, and travel. Demographic disparities, such as age, race, and gender, can also influence access to care, with certain groups experiencing barriers high enough to make care inaccessible.

1 in 2 of rural rare disease patients
traveled for more than an hour to receive care, compared to 18% of urban respondents.

3 in 10 rare disease patients
delayed care in the past year because they could not afford the visit.

1 in 4 of BIPOC rare disease patients
delayed or skipped care due to a lack of basic resources like food and shelter, compared to 1 in 10 of non-BIPOC patients.

(Source: NORD and RDDC, 2024)

Collaborative efforts between healthcare providers, patient advocacy groups, payers, and policymakers are essential to advancing access to meet the specific needs of rare disease communities. These efforts aim to mitigate disparities and improve outcomes by enhancing access to specialized care and reducing barriers to treatment across all U.S. regions. For instance, patient advocacy groups can provide crucial support and resources to patients and their families, healthcare providers and payers can work to streamline referral processes and reduce wait times, and policymakers can enact legislation that supports rare disease research and ensures that treatments are affordable and accessible. By coordinating to address the factors that influence access, we can improve the lives of those living with rare diseases and ensure that they receive the care they need.

Rare disease experts agreed the following policy actions would have significant immediate benefit as it relates to patient access to specialists and treatments.

[2](NORD - National Organization for Rare Disorders, 2022)

SOLUTION I.

Alleviate the shortage of genetic specialists by addressing licensure requirements and telehealth service coverage.

Why is this needed?

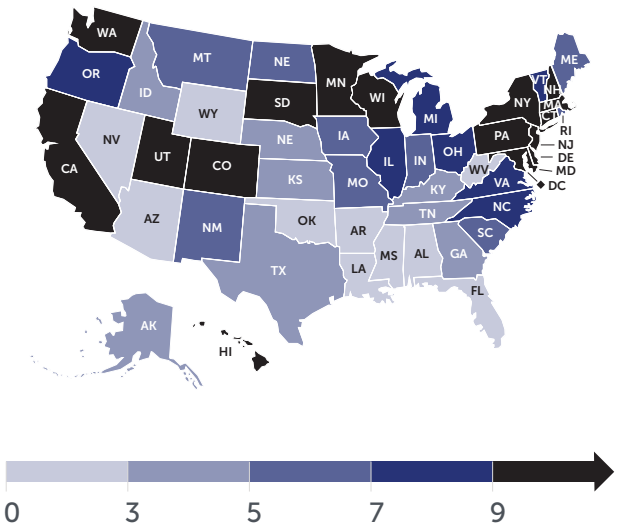
Most rare diseases have a genetic cause[3], making the role of geneticists vital in diagnosing and managing these conditions. Access to qualified geneticists and genetic counselors is crucial for the accurate diagnosis and management of most rare diseases. However, several barriers currently limit patient access to these health care providers.

Firstly, geneticists are commonly the doctors who diagnose patients with rare diseases, and the average time to receive an accurate diagnosis for a rare disease can be about 4-5 years, sometimes much longer[4]. There is a significant shortage of geneticists in the United States, and many rural areas lack these specialists entirely[5]. This shortage leads to long wait times and limited access, particularly for patients in underserved or remote areas of the country. Expanding telehealth services and ensuring all payers consistently reimburse them at the same rate as in-person visits will improve access to geneticists and testing for patients in remote or rural areas. Genetic counselors are currently not reimbursed through Medicare, nor consistently reimbursed across state Medicaid programs[6], which further reduces access to essential diagnostic services for rare diseases. As of June 2024, 34 states required genetic counseling licensure[7].

Allowing genetic counselors and geneticists to practice across state lines can further alleviate the shortage of specialists. While there are pathways to practice across statelines, most current state licensure requirements restrict the ability of these professionals to provide services to patients in other states, even via telehealth, limiting access to care[8].

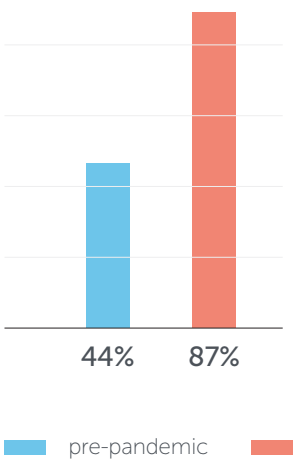
Distribution of Genetic Counselors per 500,000 People by State, 2019

As of April 2020, there were around 1,240 certified medical geneticists in the U.S., an average of 2 per 500,000 people per state.

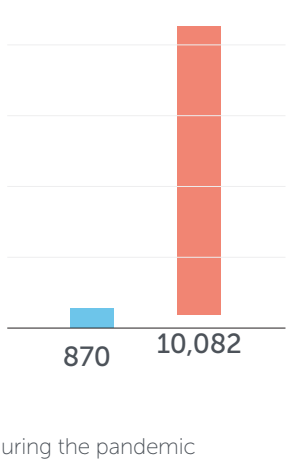


(Source: GOA, 2020)

Percentage of Genetic Counselors Using Telehealth



Telehealth Services Provided via Regional Genetics Networks-supported Clinics



(Source: NSGC, 2021)

[3](National Human Genome Research Institute, 2022)
[4](Marwaha et al., 2022)
[5](Raspa et al., 2021)
[6](NSGC - National Society of Genetic Counselors, 2024)
[7](NSGC, 2024)
[8](HHS - United States Department of Health and Human Services, 2024)

Who should act?

These stakeholders are essential to improving access to genetic specialists and telehealth services:

State Legislatures can enable improved access to genetic counselors and geneticists by allowing reciprocity of licensure across state lines.

Federal Policymakers can enact legislation to adapt reimbursement policies for telehealth services by recognizing genetic counselors as healthcare providers and ensuring telehealth services are reimbursed at parity with in-person visits.

Public and Private Insurers can ensure there are sufficient genetic specialists in their coverage networks, and provide adequate reimbursement and telehealth services so that patients have access to the necessary expertise for accurate diagnosis and management of rare diseases.

How can we measure improvement?

Tracking the reduction in geographic and socioeconomic inequities will provide insights into the effectiveness of these initiatives. This involves monitoring access to genetic specialists and telehealth services across different U.S. regions and assessing whether improvements lead to more equitable access.

Good practices for implementation

During the COVID-19 pandemic, telehealth services and reimbursements were significantly expanded to ensure continued access to healthcare while minimizing the risk of virus transmission. Medicare, Medicaid, and private insurers began reimbursing telehealth services at rates comparable to in-person visits, and geographic and site restrictions were lifted. Telehealth use went from less than one percent of visits pre-pandemic to as much as 80 percent in some areas in the spring of 2020[9]. Nearly every state modified their licensure requirements to support better access to out of state physicians during the pandemic, but those modifications were temporary and patchwork in nature[10]. When the public health emergency ended, many of these temporary measures were rolled back, but they provided a strong blueprint for good practices in this area that could be reinstated.

"For equity purposes, I think access to appropriate specialists and diagnostic tests, genetic or otherwise, is absolutely critical. One way to address that bottleneck is via telehealth."
- Michael Devlin

Several current initiatives have the potential to align licensure and reimbursement policies with the needs of patients and providers. For example, the National Society of Genetic Counselors (NSGC) advocates for standardized licensure requirements and CMS's recognition of genetic counselors as healthcare providers[11]. NORD's Rare Disease Advisory Councils (RDAC) facilitate dialogue between state legislatures and patients, which could further bring this issue to legislators' attention[12].

The Interstate Medical Licensure Compact (IMLC) simplifies the process for physicians to obtain licensure in multiple states, promoting greater flexibility and access to care across state lines[13]. Florida and Arizona have specific registration requirements that enable out-of-state physicians to provide telehealth services to patients within their borders[14]. Additional states, especially those lacking in genetic specialists, should consider implementing similar programs to enhance access.

[9](NIH, 2023)
[10](Federation of State Medical Boards, 2023)
[11](NSGC, 2024)
[12](NORD, 2024)
[13](IMLC - Interstate Medical Licensure Compact, 2021)
[14](HHS, 2024)

SOLUTION II.

Expand coverage decisions by addressing evidence gaps with strengthened post-market surveillance feedback loops.

Why is this needed?

The gap between evidence generation and coverage for rare disease treatments often leads to coverage denials, limiting patient access to necessary therapies.

Insurers have evidence standards for approving coverage which can be impossible to meet in rare diseases, where patient populations are, by definition, very small compared to populations with non-rare diseases. Even when treatments have been widely accepted by the medical community, Medicaid programs and private insurers may still classify them as experimental and not cover them due to insufficient evidence, while data to support an insurers' evidence threshold is unlikely to ever become available. For example, Luxturna, a gene therapy treatment for inherited retinal dystrophy, holds great promise for affected patients, as this condition can lead to progressive vision loss and, in some cases, blindness[15]. Luxturna was the first gene therapy for a genetic disease to be approved by the Food and Drug Administration (FDA) in 2017, and has widespread acceptance among the medical community[16].Despite that, some Medicaid programs and private insurers in 2024 still classify the drug as experimental due to the limited long-term data available[17].

Access to innovative therapies with the potential to change the lives of rare disease patients and families may be delayed significantly as these therapies undergo extended evaluation. About 80 million Americans are enrolled in Medicaid and CHIP as of June 2024[18]. With limited funds, these programs may be more inclined to cover less expensive alternatives for rare disease patients, even if they are less effective or carry more side effects. This problem is exacerbated by the fact that most rare diseases do not have a targeted therapy on the market, and for those that do, a less expensive alternative is often significantly different in efficacy.

Today's treatments for rare conditions would have been considered science fiction when I was in pharmacy school. And yet, despite that innovation, we're trying to help people living with rare conditions access these treatments with the same reimbursement system that was built for blood pressure medicines and cholesterol drugs used by millions of people. **– John O'Brien**

Rare disease patients being excluded from coverage can mean that they are forced to forego effective treatments or pay out-of-pocket for care, which can be prohibitively expensive. This lack of access can exacerbate health disparities, particularly among low-income populations who are disproportionately represented in the Medicaid system.[19]

Enhancing post-market surveillance, including patient reported outcomes when making coverage decisions, and fostering collaboration among stakeholders can help bridge this evidence vs. coverage gap and improve access to treatments.

The Friedreich's Ataxia Research Alliance (FARA) is a national non-profit advocacy organization dedicated to the pursuit of scientific research leading to treatments for Friedreich's ataxia (FA). FA is a genetic, progressive neuromuscular disease that causes ataxia, or loss of coordinated movement of the limbs. Although research and awareness of the disease often focus on the neurological symptoms, FA is multisystemic and additionally causes fatigue, scoliosis, diabetes, hearing and vision loss, and serious heart conditions. The first and only approved treatment for FA, omaveloxolone, slows progression of neurological symptoms, but its effects on FA cardiac disease have not been studied.

"Patients want to know what impact, whether positive or negative, omaveloxolone has on their heart. A post market study to understand impact on cardiac and other non-neurological aspects of FA would be meaningful to the patient community." **– Kellyn Madden**

[15](FDA - Federal Drug Administration, 2017)
[16](FDA, 2017)
[17](FDA, 2017)
[18](KFF News, 2024)
[19](Gimenez-Lozano et al., 2022)

Who should act?

These groups are essential in restoring, maintaining and enhancing incentives for rare disease drug development:

Patient Advocacy Groups are heavily involved in research and policymaking efforts for rare diseases. Organizations like the National Organization for Rare Disorders (NORD) work to advance rare disease research to support the development of effective treatments and cures, address gaps in knowledge, and advocate for favorable policies to ensure equitable access to care[20].

Public and Private Insurers should use real-world evidence and post-market data to assess the cost-effectiveness and therapeutic benefits of treatments beyond the clinical trial stage, and improve their coverage decisions and adjust coverage rulings based on that data to improve patient access to necessary therapies. They could also open up a dialogue with biopharmaceutical manufacturers earlier in clinical development of a drug to ensure that the meaningful data they need is collected during the trial, further enhancing the market approval decision-making process.

How can we measure improvement?

Monitoring the frequency of and reasons for denials can help identify areas for policy and practice enhancements. Improvement can also be measured by the implementation of regular surveys and qualitative interviews to gather patient input on their experiences and satisfaction with coverage decisions. Similarly, the number of patient representatives on review boards could be a means of tracking whether patient voices are heard. To that end, NORD launched the Living Rare Study, a longitudinal, observational study, in October 2024 designed to collect patient experience data regarding their health care experiences, resources and time needed to manage care, and disease burden on their emotional, physical, and social health[21]. This research is designed to expand our understanding of how patient experiences evolve over time and how factors, such as insurance coverage, contribute to these experiences.

Good practices for implementation

Existing U.S. programs could be expanded to further alleviate this issue. For instance, FDA's Sentinel Initiative is a national electronic system that monitors the safety of FDA-regulated medical products using data from electronic health records (EHRs), insurance claims, and patient registries[22].

Looking to international examples, Canada's Marketed Health Products Directorate (MHPD), which oversees the post-market surveillance of drugs and medical devices, collaborates with international regulatory agencies, such as the FDA and the European Medicines Agency (EMA), to enhance its ability to collect and analyze real world evidence.[23]

[20](NORD, 2024)
[21](NORD Living Rare Study, 2024)
[22](FDA, 2024)
[23](Government of Canada, 2022)

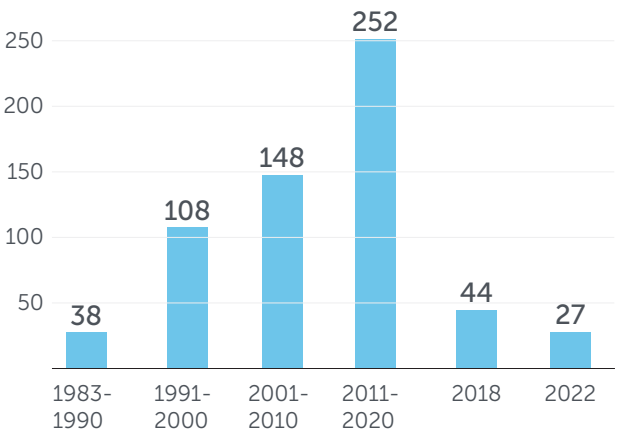
SOLUTION III.

Restore and enhance incentives to develop rare disease treatments.

Why is this needed?

The Orphan Drug Act of 1983 was designed to offer compelling incentives for the development of rare disease treatments and has led to more than 880 treatment approvals since it was enacted[24]. Unfortunately, changes in policy and economic pressures over time have reduced the scale of some of the incentives, which initially included significant tax credits, grants, and market exclusivity. For instance, the tax credit for clinical research costs was reduced from 50 to 25 percent in 2017.[25]

Orphan drugs approved over time



(Source: OJRD, 2023)

Incentives to research and develop treatments for rare disease were further diminished by the Inflation Reduction Act (IRA) of 2022, which aims to lower prescription drug costs by allowing Medicare to negotiate prices for certain medications. To implement the price negotiation piece of the IRA, the Centers for Medicare & Medicaid Services (CMS) created the Medicare Drug Price Negotiation Program (MDPNP)[26]. While the MDPNP exempts some orphan drugs from these price negotiations, this exemption is narrow. If at any time an orphan drug receives a second orphan designation or approval to treat additional conditions, it will lose its exemption to price negotiations. Because more drugs are eligible for negotiation each year in the future, this erodes the incentive to develop additional uses for a therapy, as any treatment which could provide for multiple conditions now comes with an additional financial risk. The narrow MDPNP orphan drug exemption creates risk in the already challenging arena of drug development and could disincentivize manufacturers from pursuing rare treatments.

Enhancing incentives for rare disease drug development can not only result in more rare disease drugs entering development, but can also stimulate partnerships between pharmaceutical companies, research institutions, and patient advocacy groups to meet the unmet needs of rare disease patients, specifically those currently living without an FDA-approved treatment.

30 million Americans
(~10%) living with rare disease

10,000 identified rare diseases
for which we know the molecular cause

only about 500
(less than 5%) have FDA-approved treatments

(Source: NIH, 2024)

More helpful data: https://www.saveraretreatments.org/wp-content/uploads/2024/01/Task-Force-Branded-ORPHAN-Cures-One-Pager-Revised-1.pdf

[24](Roberts, 2022)
[25](United States Congress, 2017)
[26](CMS - Centers for Medicare & Medicaid Services, 2024)

Who should act?

These groups are essential in restoring, maintaining and enhancing incentives for rare disease drug development:

Pharmaceutical Companies are key players in developing and bringing new treatments to market. Enhanced incentives will mitigate the risk of investing in rare disease treatment research, encouraging continued efforts to bring drugs to market for the thousands of rare diseases with no approved treatment.

Federal policymakers and regulators who say they support patients with rare disease have an opportunity to show their support through policies that support continued innovation and improved access to innovative treatments. – **Josh Trent**

Congressional Policymakers who want to support patients with rare diseases should take actions to ensure they have access to treatments for their disease. They must enact laws that provide financial support and regulatory flexibility, and correct the unintended consequence of the overly narrow orphan drug exclusion, to ensure orphan drug development is financially feasible and robust for the sake of people living with rare disease.

The Food and Drug Administration (FDA) plays a crucial role in administering designations and exclusivity, as well as determining if the scientific standards have been met. The FDA can ensure clarity and predictability in clinical trial design, by providing clear guidance on elements such as the use of surrogate, primary, and secondary endpoints, thereby facilitating the development process.

Patient Advocacy Groups play a vital role in using their voice to advocate for better policies and support the development of new rare disease treatments. These organizations can raise awareness, influence policy decisions, and ensure that the needs and challenges of rare disease patients are heard and addressed by policymakers and other stakeholders.

How can we measure improvement?

The number of new orphan drugs approved annually and the number of new laws to address orphan drug approval and exclusivity issues that exist today should serve as a key metric for success.

Tracking the approval rates and the diversity of conditions addressed can provide further insights into the effectiveness of incentive programs.

Good practices for implementation

In the United States, the Orphan Drug Act of 1983[27] is a notable standout for its success in fostering the development of treatments for rare diseases and highlights the importance of robust incentive structures to address unmet needs. The Creating Hope Reauthorization Act[28] which extends the Rare Pediatric Disease Priority Review Voucher program, is another example of effective policy which should be maintained and expanded. Additionally, the ORPHAN Cures Act[29], introduced in September 2023, is a bipartisan piece of legislation aimed at protecting and enhancing incentives for orphan drug development.

This act ensures that orphan drugs treating one or more rare diseases are excluded from Medicare price negotiations, as outlined in the Inflation Reduction Act, which helps maintain the financial feasibility and robustness of orphan drug development for people living with rare diseases. Addressing the orphan drug exclusion even more directly is essential to ensure that these critical treatments remain accessible and continue to receive the necessary support for development.

[27](United States Congress, 1981)
[28](United States Congress, 2024)
[29](United States Congress, 2023)

Payment Options and Prior Authorization

The economic burden of rare disease is substantial, with avoidable costs due to delayed diagnosis and treatment interruptions ranging between $86,000 and $517,000 per patient per year, according to the EveryLife Foundation for Rare Diseases[30]. Compounding this issue, employers and insurance companies are increasingly shifting the cost of healthcare onto patients by raising premiums and deductibles, further complicating access to necessary treatments.

In 2019, the total economic burden of rare diseases in the United States was estimated at **$997 billion**, with **$38 billion** in healthcare costs not covered by insurance.

(OJRD, 2019).

These financial pressures are exacerbated by the implementation of prior authorization and step therapy protocols. While these protocols are designed to control costs and prevent the overuse of healthcare services, they often inadvertently fail to consider the unique medical situations and histories of rare disease patients, including by requiring patients to repeat therapies that they have previously failed simply because of a change in coverage policy. As a result, these protocols can delay necessary treatment by weeks or even months, leading to additional financial burden on patients and potentially poorer health outcomes. To address these issues, health systems need to reexamine existing funding models and coverage structures to ensure that people living with rare diseases can access quality care and treatment.

Rare disease experts recommended the following policy actions for significant immediate benefit as it relates to patient payment options and prior authorization.

[30](EveryLife Foundation for Rare Diseases, 2023)

SOLUTION I.

Improve cost analysis approaches and implement flexible payment models.

Why is this needed?

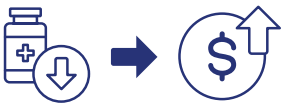
The economic burden of rare diseases is approximately 10 times higher than that of common diseases on a per patient per year basis, according to a 2022 Chiesi report[31]. Implementing flexible payment models can help manage these costs more effectively.

Currently, the lack of standardized cost-analysis approaches results in varied pricing and reimbursement decisions. This inconsistency can delay or deny patient access to essential therapies. The dominance of three pharmacy benefit managers controlling 89 percent of the market[32] further complicates access and affordability, leading to higher drug prices and out-of-pocket costs.

"I think it's really clear how broken the system is for a rare disease community and how desperate the need is to try to resolve some of these huge systematic issues." – **Heidi Ross**

Inefficient payment mechanisms do not adequately address the unique challenges of rare disease treatments. Alternative payment models that incorporate patient-reported outcome measures (PROMs) and consider costs more broadly can align treatment costs with patient outcomes, ensuring that payments reflect the real-world value provided to not only patients, but also families, carers, and society as a whole.

Value should be comprehensively defined, providing greater flexibility and a wider array of tools to address the unique needs and burdens of rare disease patients.



A lack of treatment
for a rare disease is associated with a 21.2% increase in total costs per patient per year.

(Source: Chiesi, 2022)

[31](Chiesi, 2022)

[32](FTC - Federal Trade Commission, 2024)

Who should act?

These groups are essential in designing, implementing, and managing payment models that can improve access to necessary treatments:

Public and Private Insurers can adopt and implement innovative payment models. Their collaboration is essential to ensure that these models are effectively integrated into the healthcare system. These models could be designed to capture and account for the nuances in treatment for different diseases and patient populations, ensuring that all patients are able to receive effective care.

Manufacturers play a crucial role, often collecting resource-intensive data to demonstrate the effectiveness of their treatments, ensuring regulatory compliance, and shouldering the risk of having to provide refunds or rebates if the treatment does not achieve agreed-upon outcomes. Their involvement is essential for the successful implementation and management of these models.

CMS **The Centers for Medicare & Medicaid Services (CMS)** can lead the way in setting reimbursement policies that support value-based or outcomes-based payment models. Additionally, CMS's Cell and Gene Therapy (CGT) Access Model aims to improve access to gene therapy treatments for rare disease patients with Medicaid by supporting outcomes-based agreements between states and manufacturers.[33]

How can we measure improvement?

Assessing the alignment of treatment costs with patient-reported outcomes can be measured using flexible pricing metrics and patient satisfaction surveys, which assess the alignment between the benefit of the treatment and the patient's ability to shoulder its cost.

Monitoring the reduction in out-of-pocket costs and the increase in the number of patients receiving timely treatments can provide valuable data on the effectiveness of these more flexible models.

Good practices for implementation

The Center for Medicare and Medicaid Innovation (CMMI)[34] has developed and tested numerous Alternative Payment Models (APMs) aimed at transitioning from fee-for-service to value-based care. The CMS's AHEAD Model[35] which launched in 2024 and incentivizes states to redesign healthcare delivery systems and improve equitable access to care, is another notable initiative that can serve as a blueprint for future policy development. Various healthcare stakeholders, including policy experts, healthcare providers, and patient advocacy groups, have also advocated for a broader definition of what constitutes a "value-based agreement." A broader definition would allow for the inclusion of proxy measures and patient-reported outcomes, in addition to clinically reported outcomes, thereby providing a more comprehensive understanding of value. As CMMI advances its Access Model for Cell and Gene Therapy (CGT)[36] it is crucial to include more opportunities for patient perspectives to be included to accurately collect and integrate the patient perspective.

In Canada, the integration of PROMs into value-based care models has yielded significant improvements in healthcare outcomes. By incorporating PROMs, Canada's health system has reported a 40 percent reduction in emergency room visits, a 35 percent improvement in chronic disease management outcomes, and 80 percent of patients feeling more involved in their care decisions. The use of PROMs across Canada varies by province and territory, reflecting differences in healthcare delivery models[37] However, national initiatives, such as those led by the Canadian Institute for Health Information (CIHI), have been pivotal in standardizing PROMs data collection and reporting. For instance, CIHI's national PROMs program for hip and knee arthroplasty has established guidelines for survey time points, minimum data sets, and PROMs instruments[38]. This initiative has demonstrated the importance of stakeholder engagement, including administrators, clinic managers, patients, and health system decision-makers, in achieving successful implementation.

[33](CMS, 2024)
[34](CMMI - Center for Medicare and Medicaid Innovation, 2024)
[35](CMS, 2024)
[36](CMS, 2024)
[37](Terner et al, 2021)
[38](CIHI - Canadian Institute for Health Information, 2024)

SOLUTION II.

Implement peer-to-peer review processes involving true medical peers.

Why is this needed?

Peer-to-peer reviews are conducted on a set schedule, even for lifelong medications, to provide a second opinion on the appropriateness of treatments and ensure that high standards of care and current clinical guidelines are upheld. However, in the context of rare diseases, the availability of specialists who can provide a truly qualified opinion is often limited. This scarcity can lead to authorization decisions being made without the necessary expertise, resulting in inappropriate denials and delays in care.

Delays and denials in treatment exacerbate the already substantial burden of rare diseases by leading to more severe health complications and increased healthcare utilization[39]. Patients with rare diseases are hospitalized three times more frequently than the general population[40]. People with rare diseases also often experience prolonged diagnostic journeys, which can result in progressive, irreversible, and costly complications.

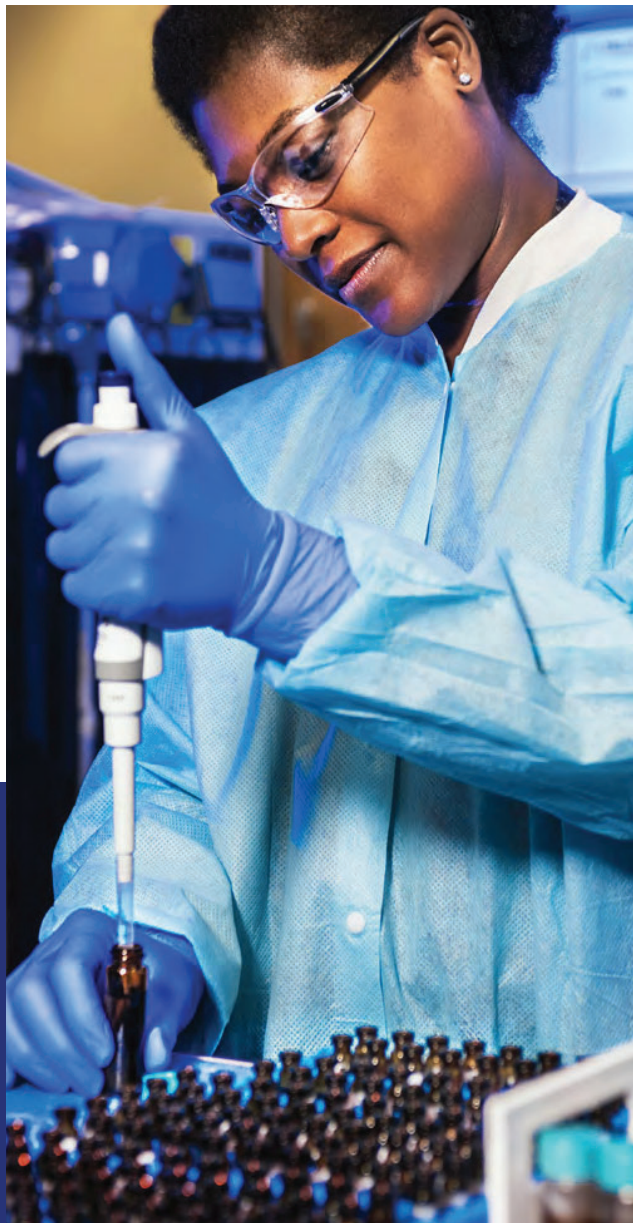
61% of patients
had been denied or faced delays accessing treatments that required pre-approval from an insurance company.
(Source: NORD, 2020)

18% of patients
had been denied referral to a specialist.
(Source: NORD, 2020)

2X
Respondents earning less than $20,000 per year were twice as likely to be denied referral to a specialist than those earning $100,000 or more.

(Source: NORD, 2020)

28% of physicians
reported that prior authorization had led to a serious adverse event for a patient in their care.
(Source: AMA, 2024)



[39](Adachi et al, 2023)
[40](NIH, 2021)

Who should act?

These groups are essential in implementing effective peer-to-peer review processes:

Public and Private Insurers need to make certain that their authorization processes are efficient and involve specialists with relevant expertise. This can help reduce inappropriate denials and improve patient outcomes.

State and Federal Policymakers are crucial in establishing frameworks and regulations that facilitate smoother and more accountable peer-to-peer review processes. Initiatives like gold carding, which allows providers with a history of high approval rates to bypass prior authorization for certain services, can significantly streamline the process and reduce administrative burdens.

Healthcare Providers, especially geneticists, play a crucial role in providing expert opinions on the necessity of treatments, particularly treatments for rare and ultra-rare diseases. Their involvement is vital to ensuring that authorization decisions are based on accurate and up-to-date clinical knowledge. Given the burden of this work, it is essential more geneticists are trained and brought into the field. In addition, considering that many rare diseases are managed by non-genetics specialists such as neurologists, immunologists, or oncologists, it is important to have increased training in and exposure to rare diseases for all clinicians, regardless of specialty. This broader training will ensure that all healthcare providers are better equipped to recognize, diagnose, and manage rare diseases, ultimately improving patient outcomes.

Instead of an adult ER doctor trying to tell me what I need for my infant with a rare disease that causes heart disease, talking to a peer [in genetics] would be really helpful, but that's hard to get. **- Christina Grant**

How can we measure improvement?

Improvement can be measured by tracking how many patients require peer-to-peer evaluation for the same therapy, the time taken to complete authorization processes, and patient outcome improvements. It's also important to note that peer-to-peer evalautions are often not conducted by a subject matter peer (i.e. a fellow geneticist), and the lack of comparable expertise can cause delays in patient approvals and impact patient outcomes. Currently, approximately 20 percent of patients may require peer-to-peer evaluation and around 80 percent of patients on lifelong medication have undergone more than one peer-to-peer process. Metrics such as reduced denial rates, faster approval times, and improved patient health outcomes can indicate the effectiveness of peer-to-peer review processes.

Good practices for implementation

The CMS Interoperability and Prior Authorization Final Rule aims to advance interoperability and improve prior authorization processes (excluding those for prescription drugs), with reports indicating a 20 percent reduction in administrative time for providers[41]. A continuous effort to expand health plan exceptions to step therapy protocols will be essential.

Turning to international models of good practice, the Canadian Medical Protective Association (CMPA)[42] supports peer review processes to ensure that authorization decisions are based on relevant clinical expertise. When organizing review panels, the CMPA carefully matches the expertise of the reviewers with the clinical specialty of the physician under review. This matching process is crucial to ensure that the review is both relevant and credible. The CMPA provides training and detailed guidelines to peer reviewers to maintain consistency and objectivity in the review process. After the review, the CMPA collects feedback from both the reviewers and the physicians being reviewed. This feedback is used to continuously improve the peer review process, ensuring it remains effective and fair.

[41](CMS, 2024)
[42](CMPA - Canadian Medical Protective Association, 2021)

SOLUTION III.

Ensure patients have continuous access to rare disease treatments by reforming utilization management, step therapy, and prior authorization.

Why is this needed?

Ensuring continuous access to necessary treatments is crucial for managing progressive diseases, as interruptions can significantly impact both disease progression and patient experience.

Patients often require new prior authorizations when changing employment or insurance, leading to interruptions in treatment. This is particularly problematic for rare disease patients who rely on continuous access to specialized treatments. Disruptions can lead to significant, and often irreversible, health setbacks, accelerating disease progression and increasing the risk of durable degradation in their condition.

43% of rare disease patients
experienced disruptions in their care due to changes in insurance or employment.

30% of patients
reported difficulties in accessing their medications after changing insurance.

25% of patients
faced increased out-of-pocket costs due to insurance changes .

Patients who experienced disruptions in their care due to insurance changes reported a 20% decrease in overall satisfaction with their healthcare. 15% of patients had to delay or skip treatments because of insurance issues.

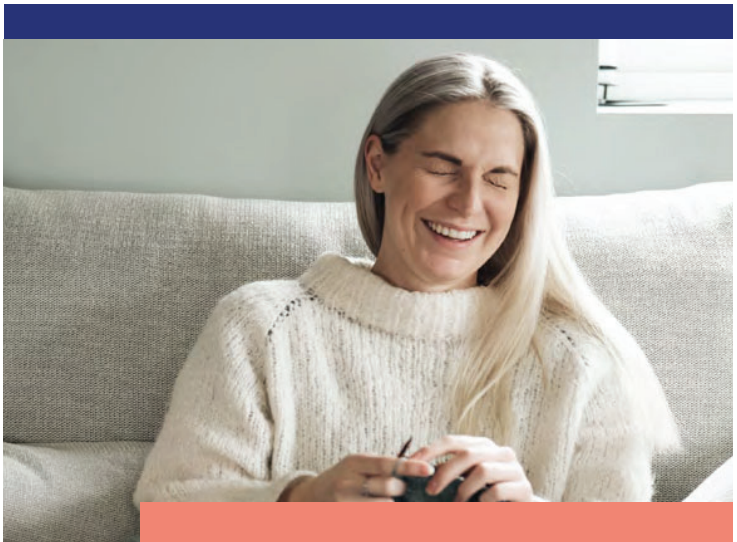
(Source: NORD and RDDC, 2024)

Ensuring that previous prior authorization approvals are maintained despite changes in insurance or employment can improve patient outcomes by preventing treatment delays that exacerbate disease progression. This continuity of care reduces the stress and uncertainty associated with treatment interruptions, thereby supporting both the physical and emotional well-being of patients.

We help patients [navigate prior authorization], we just have to do it over and over again. All the different rare diseases are doing it over and over again and facing the same exact issues. **- Kellyn Madden**

In addition to prior authorization, step therapy is a major issue on the minds of patients who can be mandated to use and fail multiple treatment options before getting the therapy originally prescribed by their doctor, causing delays and health complications. These delays are not just administrative burdens but can lead to significant setbacks in managing progressive diseases.

A comprehensive approach is needed to ensure the durability of prior authorizations and addressing the broader issues of utilization management, including step therapy. Policies should be expanded to include timely decisions around prior authorization and clear exemptions to step therapy requirements for rare disease patients.



Who should act?

These groups are essential in developing and implementing policies to ensure the durability of prior authorizations and reforming utilization management procedures:

Federal and State Policymakers can enact regulations to mandate the portability of prior authorizations across employment and insurance changes. This includes creating laws that require insurers to honor prior authorizations from previous insurers, reducing the administrative burden on patients and healthcare providers.

Public and Private Insurers need to adopt policies that recognize prior authorizations from previous insurers and implement clear exemptions to step therapy protocols to prevent unnecessary delays or gaps in treatment. Model legislation from the SAIM Coalition includes key provisions for step therapy overrides, which are also encompassed in the federal Safe Step Act[43]. According to this legislation, insurers must base step therapy protocols on independent clinical practice guidelines that are then disclosed to patients and providers so they have a clear and timely process to request an override. Patients and providers must also have a transparent and accessible appeals process, and be automatically exempt from step therapy if they are stable on their current medication, if the required medication is contraindicated, or if the patient has already tried and failed on the required medication.

CMS **The Centers for Medicare & Medicaid Services (CMS)** can lead the way as a public insurer in setting regulations that support the durability of prior authorizations. Their involvement is crucial in ensuring that these policies are adopted widely and consistently. CMS can also provide guidance and support to state-regulated insurance programs, leveraging the efforts of groups like the SAIM coalition to promote utilization management reforms at the state level.[44]

How can we measure improvement?

Monitoring the continuity of care and the number of patients who experience uninterrupted treatment despite changes in employment or insurance will help assess the impact of these policies. Surveys and health outcome data can be used to gauge patient satisfaction and the overall health impact of maintaining prior authorizations, including tracking improvements in health conditions and reductions in treatment delays. Additionally, measuring how many lives are covered by policies that minimize the prior authorization or step therapy burden will help determine the reach and effectiveness of these reforms.

Good practices for implementation

The CMS 2024 Medicare Advantage and Part D Final Rule includes provisions for maintaining prior authorizations during plan transitions, but could be further bolstered to ensure more comprehensive coverage and continuity of care.[45]

Additionally, the efforts of the SAIM coalition related to state-regulated insurance should be highlighted as a model for broader implementation, demonstrating how targeted policy changes can significantly improve patient access to necessary treatments[46]. The coalition works with state legislatures to pass laws for common-sense limits on step therapy and ensuring clear timelines for insurer responses to requests for exceptions[47].

They also focus on policies that require timely decisions on prior authorization requests and mandate the portability of prior authorizations across different insurers and employment changes to ensure that patients do not experience interruptions in their treatment due to bureaucratic hurdles.

Internationally, Australia's Medicare system includes provisions for maintaining prior authorizations during changes in insurance coverage, ensuring that patients do not face interruptions in their treatment[48]. The Canadian healthcare system supports the portability of prior authorizations across provincial borders so that patients maintain access to necessary treatments during transitions.[49]

[43](United States Congress, 2023)
[44](CMS, 2023)
[45](CMS, 2024)
[46](SAIM - State Access to Innovative Medicines, 2024)
[47](SAIM, 2024)
[48](Dixit, Sambasivan, 2018)
[49](Government of Canada, 2024)

Our Experts

The recommendations in this report were conceptualized by the U.S. experts listed below from across the rare disease landscape. These experts met in-person in September 2024 to outline policies that could be prioritized for a positive, immediate impact on the national rare disease landscape.

The recommendations presented in this report reflect the collective insights shared by the expert panel during their discussions. These recommendations are not to be interpreted as an endorsement of specific recommendations by individual panelists or the organizations they are affiliated with.

Chair: Heidi Ross
Vice President of Policy and Regulatory Affairs at National Organization for Rare Disorders (NORD)

John O'Brien
President and Chief Executive Officer, National Pharmaceutical Council (NPC)

Michael Devlin
Board Director/Strategic Advisor at the Sumaira Foundation and former Senior Vice President at Johnson & Johnson

Matt Salo
Founder and CEO of Salo Health Strategies and former founding Executive Director of the National Association of Medicaid Directors (NAMD)

Christina Grant
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