SIGNATURE SEMINARS:

WHAT'S NEXT IN RARE DISEASE

KEY INSIGHTS REPORT DECEMBER 2024

allhealthpolicy.org

ALLIANCE FOR HEALTH POLICY

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ACKNOWLEDGMENTS

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I. BACKGROUND

Summary

From September 2024 through November 2024, the Alliance for Health Policy conducted 11 listening sessions with individuals and, in some cases, pairs of participants to inform the development of rare disease seminars in fall 2024. This memo summarizes findings on the most pressing gaps and priorities on rare disease policy, as well as areas of opportunity for future bipartisan discussion.

Design and Methods Description

Participants were chosen to represent patient advocacy, government, nonprofits, and the private sector based on a set of criteria that prioritized representation of 1) bipartisan, multi-stakeholder perspectives; 2) visionary leaders driving innovation; 3) policy and political expertise; and 4) understanding of lived experience of those living with rare diseases. The interviewees were drawn from the five groups represnted below:

Listening sessions were 30-60 minutes long and conducted via Zoom. Each session followed a semi-structured interview format, including a brief introduction to the Alliance and an overview of our vision for this educational programming, along with a series of questions to better understand approaches and best practices for filling gaps in rare disease policy.

These findings are qualitative in nature and provide directional insights.

II. TOPLINE TAKEAWAYS

EXPERTS INTERVIEWED IDENTIFIED THE FOLLOWING CHALLENGES AND OPPORTUNITIES:

Challenges

1) Need to support leaps in innovation and address unmet needs: Advancing research and innovation for the many rare disease treatments needed is essential. To achieve this, strategic policy incentives are necessary to encourage development, even though biotech companies and investors face significant financial risks.

2) Cost, insurance, and affordability: There's an inherent tension between prioritizing cost-effectiveness in health care spending and the urgent need to help those with severe, life-threatening rare diseases. The financial burden of costly, innovative rare disease treatments is significant when paired with complex insurance policies and high out-of-pocket costs.

3) Creating an economic environment that addresses both the need for innovation and high cost to the system: The financial burden of costly innovative rare disease treatments is significant, compounded by complex insurance policies and high out-of-pocket costs. Balancing cost-effectiveness in health care with the pressing need to support individuals facing severe, life-threatening rare diseases is a persistent challenge. The high price of innovative treatments, compounded by intricate insurance systems and substantial out-of-pocket expenses, further complicates access to care for these vulnerable populations.

4) Burdens of time, place, and inequity on impact patients: Stakeholders frequently mention time and place as a major element of challenges in obtaining timely and affordable access to diagnosis, therapies, treatments, specialists, and other care. These include delays to diagnosis known as the "diagnostic odyssey," delays in treatment that compound the original delay, and frequent geographic barriers to required specialty care. These barriers are exacerbated for communities that are already experiencing inequity in health services, including those in minority communities.

Opportunities

1) Technologies meeting some needs of rare patients: Interviewees noted that innovations such as rapid diagnosis, personalized medicine and telehealth, and genetic testing and counseling, alongside incentivizing research and development, are paving the way for speedier diagnosis and more responsive, tailored health care solutions. Additionally, advancements in cell and gene therapy have proven transformative in some cases, offering the potential for curative solutions for some subtypes of rare diseases.

2) Regulatory efforts incentivize research and innovation: The U.S. Food and Drug Administration's accelerated approval pathway and recent establishment of the Rare Disease Innovation Hub were noted as essential steps in advancing treatment options for rare diseases.italics can be added through character styles.

What makes effective rare disease health policy?

III. KEY CHALLENGES IN RARE DISEASE

Advancing the science to address rare disease with urgency

Need to support leaps in innovation and address unmet needs: Advancing research and innovation for the diverse landscape of the thousands of rare diseases without current proven interventions is crucial. Interviewees noted that it requires strategic policy incentives to overcome the financial risks for biopharma companies and investors.

ESTABLISHING PREDICTABLE AND CLEAR INCENTIVES:

> "If you want to develop... new treatments for rare diseases, they're not financially lucrative... it costs a billion dollars to bring a drug to market... rare diseases have smaller populations for clinical trials, so the financial challenges make incentives essential." — Senior Vice President, health care Policy, Nonprofit Organization
>
> — Senior Vice President, Health Care Policy, Nonprofit Organization

- "We need incentives for companies to be looking at rare disease... development to support overall patient care and help mitigate the high costs and risks involved." — Senior Managing Director, Private Sector
- "The Orphan Drug Act and other incentives are necessary for encouraging investment in rare diseases, but companies still face challenges when incentives are too limited or regulations shift unpredictably... It's crucial to have these policies stable and clear for companies." — Vice President of State Policy, Nonprofit Organization
- "It is critical to ensure continuous encouragement for pharmaceuticals and other companies to invest in rare disease therapeutics, as these treatments are often not cost-effective under standard market conditions." — VP of Public Health and Policy, Nonprofit Organization
- "One challenge is incentivizing companies to explore additional indications for existing drugs, as current regulations sometimes discourage this by introducing tax penalties or reducing profitability for multi-indication treatments." — Chief of Policy, Advocacy & Patient Engagement, Nonprofit Organization
- "People focus on two types of incentives to get people to develop rare diseases: push incentives like making clinical trials cheaper through expedited reviews and tax credits, and pull incentives to make treatments lucrative, such as guarantees that therapies will be reimbursed or transferable expedited review vouchers... policies to protect financial viability for developing therapies in rare diseases are necessary to incentivize companies to prioritize these conditions." — Senior Managing Director, Private Sector

TREATMENT GAPS:

Significant unmet needs in rare disease treatments present unique challenges in a mismatch between the ability to diagnose and treat conditions.

- "If we screen these children, diagnose them, and there are no treatments, it raises ethical and financial challenges. Drug development is so costly and complex for these small patient populations that many treatments simply aren't available."— Director of Community Engagement, Nonprofit Organization
- "Identifying these conditions early doesn't always lead to treatment because of high development and reimbursement costs... Financing treatments in this space remains a major barrier to access." — Senior Vice President, health care Policy, Nonprofit Organization

Insurance and affordability

CREATING AN ECONOMIC ENVIRONMENT THAT ADDRESSES BOTH THE NEED FOR INNOVATION AND HIGH COST TO THE SYSTEM

There's an inherent tension between prioritizing cost-effectiveness in health care spending and the urgent need to help those with severe, life-threatening rare diseases. The financial burden of costly innovative rare disease treatments is significant, compounded by complex insurance policies and high out-of-pocket costs.

- "Rare disease drugs are often very expensive per patient due to the high costs associated with their development... There's a concern about affordability on a per-person basis, even though rare disease treatments are helping the most severely affected patients." — Senior Managing Director, Private Sector
- "There's kind of this mentality in the rare disease community... they (patients) will pay any amount for a therapy if it comes to market, which is really disappointing." — Health Policy Advisor US House (D)
- "But I think particularly with rare diseases, there's a sort of an unpalatability. It's not very [palatable] to think that we're going to be making a profit off of the five children who have this terrible disease. ... Money's going to drive so much of this because these are so expensive. ... I hate to say that it's just horrifying to have money and, you know, a child's life and money intertwined. But it's a reality." — Consulting Actuary, Private Sector

INSURANCE CHALLENGES

Interviewees note that the tools that insurers use to manage higher cost treatments can represent hurdles for rare disease patients in accessing timely treatment. These include strict prior authorizations, high patient cost responsibilities, such as co-pays or premiums, and complex coverage rules that can be difficult to navigate and result in a delay or limited access to essential therapies. Such delays risk worsening health outcomes and increasing financial strain, which leads to the need that rare disease advocates highlight for streamlined insurance processes and reduced financial barriers to ensure timely and affordable access to life-saving care.

- "In the U.S., treatment delays might not be as common as in other countries, but they're still significant, especially for costly rare disease therapies. If insurance companies are cost-sharing, patients end up with hefty out-of-pocket expenses before they can even start treatment." — Senior Managing Director, Private Sector
- "Many rare disease patients have insurance, but understanding coverage is confusing... even if they have it, there's often no clear path to what's covered. It's time-consuming, and the system almost creates delays by design." — VP of Public Health and Policy, Nonprofit Organization
- "For most patients, having a treatment available doesn't mean you can access it. Rare disease treatments... are frequently expensive. And so insurance protocols often do their best to limit access. We see that through... prior authorizations that just delay or deny care." — Executive Director, Nonprofit
- "The biggest challenge is hedging risk and making sure there's money to pay for extremely expensive treatments at a reasonable price." — Consulting Actuary, Private Sector

Time, place and equity challenges

Stakeholders frequently mention time and place as major challenges in obtaining timely and affordable access to therapies, treatments, specialists, and other care. These include delays to diagnosis known as the "diagnostic odyssey," delays in treatment that compound the original delay, and frequent geographic barriers to specialty care required. These barriers are exacerbated for communities that are already experiencing inequity in health services, including those in minority communities. Furthermore, issues related to geographic barriers, race, and other attributes exacerbate these challenges.

DIAGNOSTIC DELAYS- THE "DIAGNOSTIC ODYSSEY" FOR RARE PATIENTS

Diagnostic delays are common in rare diseases, with patients frequently experiencing years of uncertainty before receiving an accurate diagnosis. Such delays contribute to disease progression and complicate treatment, underscoring the need for streamlined diagnostic pathways. Enhancing early diagnostic efforts, and improving coordination amongst providers, can improve outcomes and quality of life for rare disease patients.

> "Patients often experience delays in diagnosis due to a lack of awareness among primary care physicians and specialists. Rare diseases often involve multiple specialties, which creates a need for more efficient referral systems to specialists."
>
> — Executive Director, Nonprofit

- "Time and again, we hear from people that they've been trying to get a diagnosis for 10, 20, 30 years. In the vision world, every year that you delay getting a diagnosis is a year of vision loss... Getting somebody to diagnosis faster... prevents further vision loss, which helps everything else in their life." — VP of Public Health and Policy, Nonprofit Organization
- "There is the devastating situation of going through all that it takes to even get a rare disease diagnosis, then finding out that there's a treatment. But then the final step, the reimbursement often seems to be consistent across all of the rare diseases. So... it sort of leads to either diagnostic delays or treatment delays because of affordability aspects."— Senior Vice President, health care Policy, Nonprofit Organization
- "I think diagnostic delays due to race and ethnicity exist... With rare disease, it makes it that much more difficult because it's less front of mind for a physician who's diagnosing." — Vice President of State Policy, Nonprofit Organization

TREATMENT DELAYS

Treatment delays in rare diseases often occur due to limited treatment availability, insurance barriers, or high costs. These setbacks can worsen patient health and increase the burden on health care systems. Policies that address these delays are essential for ensuring timely access to life-saving therapies for rare disease patients.

- "For most rare diseases, you know, you may have one, maybe two treatments. Let's make sure you have access, and... that shouldn't really be an impediment, especially if people have a lot of extra costs, in terms of maybe one of the parents having to quit their job to take care of someone... delays are really a huge issue. You know, as new therapies come out, you want people to have access to them as soon as possible, especially because there's typically fewer treatment options." — Senior Managing Director, Private Sector
- "What is difficult is access to this drug or access to this type of treatment. So the disease itself is covered. The best treatment for it may be harder because of supply issues or access by distance or contracting issues or the question of it being investigational. Do you remember when every time a new super modern disease comes out, it turns out there are five people in the United States that can administer it? And if you live in rural Oregon, none of them are close." — Consulting Actuary, Private Sector

GEOGRAPHIC BARRIERS

Geographic barriers pose significant challenges for rare disease patients, often requiring travel to specialized centers located far from home. These delays can prevent timely access to essential care and treatments, disproportionately affecting patients in rural and underserved areas. Addressing these barriers is crucial for equitable access to health care services.

- "... if you're in Alaska, you're flying to Seattle... you shouldn't have to have a flight to check in with your provider if that's the closest clinic." — Vice President of State Policy, Nonprofit Organization
- "If I've been either disincentivized or [had it] made close to impossible for me to travel... then I'm not going to take advantage of medical care that is outside of driving distance. And I've seen this happen a lot." — Director of Community Engagement, Nonprofit Organization
- "Many of our conditions in the rare world... still require people to see specialists that are mostly in metropolitan areas. You aren't going to find [them] out in the frontier lands. So people are still going to have to travel, and the need in the rare disease community for access to specialists is significantly higher because they just can't have their needs met elsewhere" — VP of Public Health and Policy, Nonprofit Organization
- "The problem is that while plans are all over the country, the specialists who are knowledgeable enough to administer some of these are frequently clustered around academic centers. And it can be a long way from where the person lives to the academic center. And the plan may not have contracts with the academic center." — Consulting Actuary, Private Sector

IV. OPPORTUNITIES

Technologies that move toward a patient-centered approach

Interviewees noted that innovations such as rapid diagnosis, telehealth, and genetic testing and counseling are paving the way for speedier diagnosis and responsive, tailored health care solutions. Advancements in cell and gene therapy have proven transformative in some rare diseases, offering the potential of curative solutions for some subtypes of rare diseases. To change the text weight, set italics or change the color use the formatting options under character styles.

RAPID DIAGNOSTIC CAPABILITIES

Rapid diagnostic capabilities are essential in rare disease care, enabling timely identification of conditions that often go undiagnosed for years. Early and accurate diagnosis not only improves patient outcomes, but also helps reduce health care costs by directing patients to appropriate treatments sooner. As technology advances, enhancing diagnostic speed and accessibility is a critical focus in addressing the unique challenges of rare diseases.

- "The average time of diagnosis for, I think it's seven years, you know, for a rare disease patient, one of the ways you can speed that up and stop people from having disease progression and untimely, you know, medical events and all of that is by diagnosing earlier." — Executive Director, Nonprofit
- "For rare pediatric disorders, newborn screening is the most effective method for early identification. Rapid diagnosis can lead to better outcomes, but there's pushback because, often, treatment options are still limited, making the value of diagnosis a challenging discussion." — Senior Vice President, health care Policy, Nonprofit Organization
- "Better diagnostic tests and development of those are essential for patient care. The current system takes so long to get a diagnosis that many patients lose functionality while they wait. Faster, more accessible diagnostics would make a real difference in rare disease treatment." — VP of Public Health and Policy, Nonprofit Organization
- "There is a huge need in the rare disease community for diagnostic innovation to shorten the diagnostic odyssey. We are really focused on policies that can help ensure earlier diagnosis and access to appropriate care." — Chief of Policy, Advocacy & Patient Engagement, Nonprofit Organization

TELEHEALTH AS A STEP FORWARD

Telehealth plays a crucial role in reducing geographic and financial barriers to care for rare disease patients, particularly those who need regular follow-ups or are enrolled in a clinical trial.

- "Telehealth can be a part of clinical trials... we can have fewer travel needs in the trial process. A lot of rare disease patients face travel difficulties for clinical trials that are centralized in locations like Boston or San Francisco. If telehealth can be a part of clinical trials, we can reduce travel requirements. Stakeholders support telehealth as a way to ease logistical challenges for patients who need specialty care but live far from urban centers." — Director of Community Engagement, Nonprofit Organization
- "Being able to access telehealth to deal with some of the day-to-day is helpful." — Vice President of State Policy, Nonprofit Organization
- "We try to get at it somewhat through telehealth, but also as we see more and more folks enroll in Medicare Advantage plans, we're seeing a real failure of these networks to be adequate provider networks. And so folks aren't able to access... specialty clinics... telehealth is helpful." — Senior Managing Director, Private Sector
- "At the federal level, we have many conditions with effective screens and life-changing treatments that we are not yet screening for federally... modernizing our national system to reduce hurdles... telehealth could alleviate these gaps." — Chief of Policy, Advocacy & Patient Engagement, Nonprofit Organization

GENETIC TESTING AND COUNSELING

Genetic testing and counseling are important for the advancement of personalized medicine. Genetic testing can help diagnose diseases and determine which treatments might be useful for a patient. Concerns about privacy are at the forefront of policy concerns and must be taken into consideration as genetic testing and counseling moves forward.

> "The ability to access genetic testing and counseling is critical. The earlier patients know what they are dealing with, the better we can manage their disease journey and even prevent certain outcomes, but it requires supportive policies and insurance coverage to be truly effective."
>
> — Executive Director, Nonprofit

- "Access to genetic tests and genomic sequencing is often a critical step for children with rare disease getting an accurate diagnosis. Improving Medicaid coverage of tests has been a long-standing area of bipartisan interest for Congressional policymakers." — Managing Principal, Private Sector
- "So in terms of policy gaps, you know, we're still at a point where, it's what, 95 percent of rare diseases don't have a treatment? And so, you know, you've got a huge need for policies that will drive, you know, access to genetic testing and counseling, which play a role in the patient's disease journey. We need to pass [legislation] to ensure that people can access the types of genetic testing or biomarker testing that they need." — Executive Director, Nonprofit
- "GINA [Genetic Information Non-disclosure Act] went a long way, did some good in terms of getting insurance, but then there were types of insurance that were not included there. So you could conceivably have your genetic predisposition to a disease disqualify you from life insurance or long-term care insurance." — Director of Community Engagement, Nonprofit Organization
- "We do have privacy protections for genetic testing results, but there are still places where you can be discriminated against, such as with life insurance. Solving for these gaps is critical to ensuring access to genetic testing." — Executive Director, Nonprofit
- "Privacy concerns exist over genetic information being used for discrimination, such as life insurance denials. Policies like the Genetic Testing Protection Act in Florida aim to prevent such practices." — Vice President of State Policy, Nonprofit Organization

CELL AND GENE THERAPIES

Cell and gene therapies represent a groundbreaking shift in rare disease treatment, offering the potential for curative solutions rather than traditional symptom management. These therapies, particularly suited for single-gene mutation conditions, have already shown promise in diseases like sickle cell disease and beta-thalassemia. As the first commercialized gene therapies gain traction, they set critical precedents for innovation while highlighting challenges such as affordability, accessibility, and the need for supportive policies to ensure equitable implementation.

- "Gene and cell therapies are highlighted for their potential to cure single-mutation diseases entirely, as seen with therapies for sickle cell disease and beta-thalassemia." — Senior Managing Director, Private Sector
- "The sickle cell disease treatment was important not just for that community, but also because it was the first commercialized gene therapy in the U.S. Its success sets a critical precedent for other therapies in the pipeline... Gene therapies, while revolutionary, come with high costs and logistical barriers such as the need for extensive follow-up and monitoring, which may limit their accessibility to certain populations." — Senior Vice President, health care Policy, Nonprofit Organization
- "Proposals for more flexible emergency use authorization to address fatal and fast-progressing diseases through emerging genetic and immunotherapy technologies have been highlighted as a way to accelerate treatment availability for rare disease patients." — Legislative Assistant, US House (R)

Regulatory landscape opportunities

The FDA's accelerated approval pathway and recent establishment of the Rare Disease Innovation Hub were noted as essential steps in advancing treatment options for rare diseases. These initiatives aim to expedite the availability of therapies for conditions that lack adequate treatment by enabling faster approval of promising drugs. The accelerated approval process allows therapies to reach patients sooner by permitting market access based on preliminary evidence, while the innovation hub focuses on creating streamlined pathways for developing and approving treatments. Together, these efforts encourage pharmaceutical investment in rare disease research and aim to improve patient outcomes more swiftly and effectively.

> "The Hub has three primary functions: (1) serve as a single point of connection and engagement with the rare disease community; (2) enhance FDA collaboration to address common scientific, clinical and policy issues related to rare disease product development; (3) advance regulatory science with dedicated work streams for consideration of novel endpoints, biomarker development and assays, innovative trial design, real world evidence, and statistical methods." — Managing Principal, Private Sector
>
> — Managing Principal, Private Sector

- "The FDA has not shown itself to be historically very good at approving drugs for small patient populations... that's where a lot of the rare disease communities' attentions have focused over the past few years, like pushing FDA to really innovate and find new ways to approve new therapies... We actually have seen some progress from the FDA this year, and the FDA's Rare Disease Innovation Hub is a positive step, but there's more we can do there." — Health Policy Advisor, US House (D)
- "We're seeing some progress from the FDA with their Rare Disease Innovation Hub... They're getting more serious about addressing issues in the rare disease community, but we still have work to do." — Vice President of State Policy, Nonprofit Organization
- "The accelerated approval pathway has been really important for rare disease... We want to make sure we protect that pathway because that is how a lot of rare disease treatments get approved." — Executive Director, Nonprofit
- "We're focused on advocating for regulatory pathways that incentivize development without reducing rigor... accelerated approvals can make a meaningful impact by addressing unmet needs in rare disease." — Chief of Policy, Advocacy & Patient Engagement, Nonprofit Organization

V. CONCLUSION

Our findings show that rare disease is an area of policy with thousands of conditions and even more unmet needs. While much of the policy conversation centers on how to incentivize breakthroughs, experts note that there is ample opportunity to address additional unique challenges these conditions present to patients, policymakers, and the health care system, including holistic impacts on patients like financial, mental health, and geographic factors. According to our interviewees, sound approaches to rare disease policy should address issues of incentives, costs, and sustainability; take on the non-financial barriers to care; and fully leverage new technologies to make access to the unique services that rare conditions require easier for patients.

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