THE RIGHT CARE FOR RARE

May 2024

A POLICY ROADMAP
TO SUPPORT RARE DISEASE PATIENTS

OVERVIEW

About one in 10 people in the United States lives with a rare disease, which is a health condition that impacts fewer than 200,000 people.[1] Yet, of the more than 7,000 diseases that meet this criteria, an estimated 95% do not have an FDA-approved treatment.[2]

Rare disease patients need a clearer path to diagnosis, more treatments and swifter access to timely care. Policies must acknowledge these needs and encourage innovative treatments for people living with rare diseases.

This roadmap outlines four key ways that policymakers can support rare disease patients.

1.  EMPOWER PATIENTS
2.  REDUCE ACCESS BARRIERS
3.  CONFRONT PAYMENT ISSUES
4.  PROTECT & ENCOURAGE INNOVATION

STEP 1

EMPOWER PATIENTS

Rare disease patients can face numerous challenges as they search for a diagnosis or seek out the right treatment. Considering that the average time from symptom onset to diagnosis is six years, empowering patients throughout their rare disease journey is a necessary first step.[3]

IMPROVE ACCESS TO SPECIALISTS

Shortening the period from symptoms to diagnosis, which can take several years, is often key to patient outcomes. In many cases, one important step in getting patients to a diagnosis is connecting them with the right specialists earlier.

There is, unfortunately, low awareness for many rare diseases, and the providers best informed and prepared to diagnose a rare disease are those with specialized knowledge. But accessing a specialist can be tricky because of:

- Distance from the specialist
- Transportation costs
- Coverage restrictions
- Telehealth limitations
- A lack of specialists

Bridging the gap between patients and specialists can shorten the length of time it takes to get a diagnosis. Without access to the right specialists, patients may experience lengthy periods without a diagnosis or appropriate treatment. They may even receive a misdiagnosis because clinicians don't recognize the disease's symptoms for what they are.

Good policies can set patients up to easily and swiftly access specialists. Expanding telehealth opportunities and ensuring patients can reach specialists who may be far away are important steps. Connecting patients quickly matters. It can be the difference between months and years of searching for a diagnosis.

EXPAND GENETIC TESTING & COUNSELING

About 80% of rare disorders are genetic,[4] so genetic testing is one of the best ways to reach a diagnosis faster. Since the mapping of the human genome in 2003, more than 75,000 genetic tests have emerged, but some tests are expensive and may not be covered by insurers.[5]

After genetic testing, working with a genetic counselor can help patients understand their condition and their treatment options. However, genetic counselors are in short supply. Some patients, including those in rural areas, may have limited access to this type of clinician. Meanwhile, some insurers may cover the genetic test but not the counseling that makes the test results actionable.

Policymakers can work to ensure coverage of genetic testing and counseling. Counselors must have adequate resources and the opportunity to recommend treatment plans to clinicians.

ESTABLISH RARE DISEASE ADVISORY COUNCILS IN EVERY STATE

Advocates and policymakers can also take steps to ensure rare disease patients have a voice in state-level policies. Rare disease advisory councils are one important example of advisory bodies that give the rare disease community a stronger voice in state government.

The first rare disease advisory council was created in North Carolina in 2015. Since then, more than two dozen additional states have established councils. These organizations help states identify and address barriers that prevent people with rare diseases from obtaining proper treatment.[6]

Actively working to establish and successfully implement these councils in every state should remain a high policy priority.

During the creation of rare disease advisory councils, policymakers should prioritize:

- Ensuring clear communication between patients and policymakers
- Elevating rare disease patients' experiences and needs
- Including the patient and provider perspective
- Enhancing rare disease education and awareness

STEP 2

REDUCE ACCESS BARRIERS

More than 95% of rare disease don't have an approved treatment, but innovation is expanding options for rare disease patients. Accessing these innovative options, however, can sometimes be challenging. Coverage barriers and value discussions can all complicate a patient's treatment path.

Policymakers can take steps to help rare disease patients overcome access barriers.

LIMIT HARMFUL UTILIZATION MANAGEMENT PRACTICES

Sometimes health plans use bureaucratic tactics to reduce insurers' costs—frequently at the expense of patient access. These practices are known as utilization management.

Prior authorization is one well-known example. A clinician may prescribe a medication, but then the health plan withholds coverage until the provider submits specific forms and demonstrates that the health plan's criteria have been met. The process can drag out, especially if the health plan denies the request and the patient's provider has to appeal. In the meantime, the patient goes without treatment.

Rare disease patients typically have limited options, and prior authorizations may lead to devastating consequences. Delays in care while waiting for a treatment to be approved can lead to extended suffering and disease progression. And in some cases, getting coverage for a medication may be challenging, especially if insurers aren't familiar with the rare condition or its standard of care.

A related and commonly used utilization management tool is step therapy. Here patients are required to "fail" one or more medications, typically those that are less expensive for the insurer, before they can access their prescribed medication. Some patients must try several different medications, even as their condition progresses without adequate treatment.

Another insurance cost-cutting practice is non-medical switching, when the health plan changes a stable patient's treatment regimen to reduce their own costs or maximize their profits. In a nationwide poll of 800 patients who experienced non-medical switching firsthand, almost 40% of patients said their new medicine was not as effective as their original. Nearly 60% had complications.[7]

Patients with more than one treatment option may face non-medical switching. This tactic can impact patients' ability to remain stable, and it may even lead to additional side effects or disease progression.

LIMIT PRESCRIPTION DRUG AFFORDABILITY BOARDS' HARMFUL IMPACT

Recently, more and more state legislatures around the country have begun creating prescription drug affordability boards. These organizations seek to reduce states' spending on medications, but they may end up limiting patients' access.

Many of these boards apply a metric known as the quality-adjusted life year, which was created by economists rather than health care experts. This metric gauges medication value relative to how many years of "perfect health" it provides. The approach can diminish the value of health for patients with disabilities or chronic and incurable conditions, including rare diseases.

These boards should incorporate the perspectives of rare disease patients and providers and ensure access to rare disease medications remains intact. At a minimum, advocates must persuade state policymakers to eliminate the use of discriminatory metrics like the QALY.

STEP 3

CONFRONT PAYMENT ISSUES

Access challenges aren't the only barrier that patients face. Whether or not patients can afford their out-of-pocket cost for a treatment can sway the decision to adhere to — or abandon — medication.

RESTRICT COPAY ACCUMULATOR & COPAY MAXIMIZER PROGRAMS

Many insurance policies leverage copay accumulator programs, which prohibit copay cards and assistance from counting toward patients' deductibles or annual out-of-pocket expenses. When patients exhaust their copay assistance, they may discover that they unexpectedly owe a large amount to get their medication.

Similarly, copay maximizer programs are written into health plans when the insurer decides that certain payments made by a drug manufacturer will not count toward the patient's deductible and out-of-pocket maximums for the year. Payments don't count toward patients' deductibles, often leaving them with a greater financial burden as they try to pay for their medication and other health expenses.

Since medications that treat rare diseases are often expensive, copay accumulators and maximizers can make treatment cost prohibitive for some patients. When these patients face extreme financial choices, they may ration their medication or stop treatment altogether, which can worsen their symptoms and health outcomes.

State and federal policy successes have limited these programs. But more advocacy work must be done. Limiting or banning copay accumulators and maximizers is necessary to ensure optimal care for patients with rare diseases.

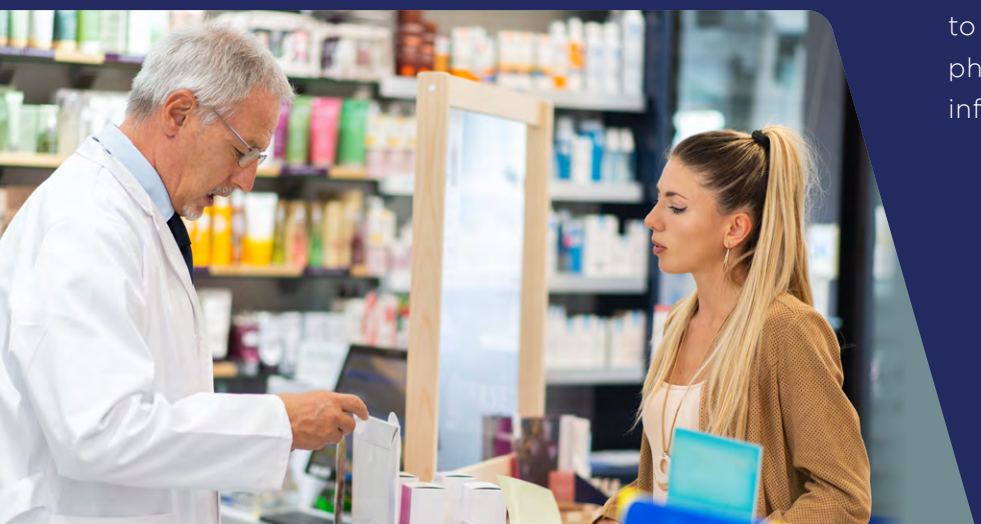
RESTRICT ALTERNATIVE FUNDING PROGRAMS

In recent years, a new threat to access and coverage for patients with rare diseases has materialized. Third-party vendors work with employers to implement alternative funding programs, which can make it harder to access the very type of specialty medications that rare disease patients often need.

Third-party vendors advise employers to remove specific high-cost drugs from their health plans' formularies by categorizing these medications as "non-essential" health benefits. Alternative funding programs then divert insured patients to patient assistance programs to get certain medications.

For patients with rare diseases, these same treatments are essential—even life-saving. But with the medications arbitrarily carved out of their health plan, insured patients with rare diseases are now effectively uninsured. Despite having insurance, they can access their medication only with the help of programs intended to help the truly needy.

Advocates and policymakers must work to restrict alternative funding programs, making sure that insured patients have the coverage they deserve.



INCREASE TRANSPARENCY FOR PHARMACY BENEFIT MANAGERS

Pharmacy benefit managers are classic middlemen, hired by insurance plans to negotiate drug prices with manufacturers and manage claims with pharmacies. Today just three pharmacy benefit managers—Express Scripts, CVS Caremark and OptumRx—control approximately 89% of the market and impact more than 270 million American lives.[8]

Pharmacy benefit managers shape what a health plan covers. They can also drive prescription drug prices up by demanding higher and higher rebates from manufacturers, resulting in higher out-of-pocket costs for patients who pay coinsurance, a percentage of their medication's cost.

Significant efforts at the federal and state level can help reform and fix the pharmacy benefit manager system. Advocates and policymakers must aim to increase transparency and limit pharmacy benefit managers' growing influence on patient access.

STEP 4

PROTECT & ENCOURAGE INNOVATION

Most rare diseases don't have a treatment yet, making policies that encourage research and development critical for patients with rare diseases.

URGE CONGRESS TO **PASS GOOD POLICIES**

The Inflation Reduction Act's drug price negotiation program may discourage innovation and investment in new treatments for rare diseases. Several bills before Congress, however, could make a positive impact for rare patients.

The ORPHAN Cures Act would preserve the orphan drug status of medications that treat one or more rare diseases, exempting them from the Medicare drug price negotiation program. Another bill, the MINI Act, would encourage ongoing research and development for genetically targeted technologies. These novel medications treat a number of rare conditions at the genetic level.

These bills would support continued innovation for rare diseases, ensuring that patients without an FDA-approved treatment can benefit from future therapies. Development of new treatments can remain a priority for manufacturers, and many rare disease patients could see treatments for their condition come down the pipeline.

ENSURE THE FDA & CONGRESS **PROTECT ACCELERATED APPROVAL PATHWAYS**

The Accelerated Approval Pathway is a crucial way for patients to access innovative medications quickly and efficiently. So few rare diseases have an FDA-approved treatment created specifically for the condition, and rare disease patients may go years searching for the right treatment. The accelerated approval pathway makes it possible for innovative treatments to quickly and effectively undergo review, ultimately reaching patients who need new options.

Advocates and policymakers should urge the FDA and Congress to protect the Accelerated Approval Pathway and patients' access to treatments approved through the pathway.

CONCLUSION

Rare disease patients face many challenges. Policymakers, however, can stand up for rare disease patients and implement policies that:

- Empower patients
- Reduce access barriers
- Address payment issues
- Protect and encourage innovation

Rare disease patients deserve timely access to accurate diagnoses and tailored treatment. Policymakers can do their part by ensuring that good policies aren't rare.

REFERENCES

1. National Institutes of Health. Genetic and Rare Diseases Information Center. https://rarediseases.info.nih.gov/about
2. National Institutes of Health. Delivering Hope for Rare Diseases. January 2023. https://ncats.nih.gov/sites/default/files/NCATS_RareDiseasesFactSheet.pdf
3. National Organization for Rare Disorders. Four actions for faster rare disease diagnosis and improved health equity, a Takeda & NORD report. October 2023. https://rarediseases.org/in-pursuit-of-health-equity/
4. The Human Genomic Research Institute. Rare Genetic Diseases. https://www.genome.gov/dna-day/15-ways/rare-genetic-diseases
5. Phillips K, Deverka P, Hooker G, Douglas M. Genetic Test Availability and Spending: Where Are We Now? Where Are We Going? Health Affairs. 2018 May; 37 (5): 710-716. DOI: 10.1377/hlthaff.2017.1427. https://www.healthaffairs.org/doi/citedby/10.1377/hlthaff.2017.1427
6. National Organization for Rare Disorders. Rare Disease Advisory Councils. https://rarediseases.org/policy-issues/rare-disease-advisory-councils/
7. Alliance for Patient Access. A Study of the Qualitative Impact of Non-Medical Switching. February 2019. https://admin.allianceforpatientaccess.org/wp-content/uploads/2020/02/AfPA_Qualitative-Impact-of-Non-Medical-Switching_Report_Feb-2019.pdf
8. National Association of Insurance Commissioners. Pharmacy Benefit Managers. 2022 April 11. https://content.naic.org/cipr-topics/pharmacy-benefit-managers

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ABOUT THE ALLIANCE FOR PATIENT ACCESS

The Alliance for Patient Access is a national network of policy-minded health care providers advocating for patient-centered care.

AllianceforPatientAccess.org