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Addressing Unmet Needs for Patients with **Rare Vision Conditions**

VISION HEALTH
ADVOCACY COALITION

Introduction

About one in 10 people in the United States lives with a rare disease, defined as a condition that impacts fewer than 200,000 people.[1] Yet 95% of rare diseases have no FDA-approved treatment.[2]

Rare diseases related to the eye account for a significant number of rare conditions. Such conditions can be disruptive and debilitating, but little public awareness exists of rare vision diseases. As a result, patients often suffer in relative silence, and barriers to treatment can lead to severe and permanent damage to the eye.

The struggle to get a diagnosis can delay timely treatment, as many general practitioners and optometrists have little or no experience with rare vision conditions. In fact, it typically takes more than five years for patients to receive a rare vision disease diagnosis.[3] The inability to access qualified medical specialists can also be a barrier, according to the American Medical Association. There are too few specialists, with a shortage projected to reach up to 77,100 specialists. Meanwhile, most practicing specialists are concentrated in high-population areas.[4]

Even after patients receive a diagnosis, still more barriers present themselves. Pinpointing these challenges is an essential first step toward crafting solutions to address them.

Barriers to Treating Rare Vision Diseases

Long Diagnostic Journeys

Diagnosing a rare disease can take years. Patients with rare diseases often have to consult primary care physicians, who are unlikely to be familiar with their condition. They are then referred to specialists, who may have a waiting list to see new patients and may take weeks or months to reach their own determination. Second or third opinions may add many more months to the process, further diminishing the probability of positive patient outcomes.

Rare diseases, including rare vision diseases, tend to result from inherited genetic anomalies. Genetic testing can give providers a closer look into patients' genes.

If a mutation is found, providers can better understand what disease is affecting their patient and can develop a personalized course of treatment.

Therefore, the quickest and most conclusive path to diagnosis typically involves genetic testing. But there are multiple obstacles to reaching a diagnosis through genetic testing. The first is awareness, on the part of both the patient and their provider.

More than 70,000 genetic tests exist, but awareness of the options is still limited, making patient and provider education critical.[5]

Policy Solutions for Long Diagnostic Journeys

Patient and provider education is an important step forward. **Patients need a clear understanding of where they will receive care, what the care will look like and how their treatment will unfold.**

Health care providers can grow in their knowledge about rare vision diseases, technology improvements and new medications coming to market. Many states now have rare disease advisory councils, which work to educate the public, medical professionals and policymakers about rare diseases. Expanding these opportunities allows for greater engagement and more inclusive policy conversations about value and access to life-changing treatment.

This is particularly important as estimates of medications' value, such as reports from health technology assessment organization the Institute for Clinical and Economic Review, influence coverage decisions at the health plan, state and even national level.

Patients also need support from family members and care partners. To support patients and caregivers, caregiver groups need to be part of the conversation and have the opportunity to disseminate information about rare vision diseases to their members. Building a community allows people with rare eyes diseases to validate and share their experiences.

Inability to Access Genetic Testing and Genetic Counseling

There is a severe shortage of medical specialists in genetic testing and counseling, especially those who specialize in rare or rare vision diseases. Patients who seek out genetic testing may have to travel long distances to complete the testing, further delaying their diagnosis.

Once patients understand their genetic testing options, they may have anxiety about learning their results.

Some may worry about how confirming a genetic anomaly could impact their employment prospects, life insurance premiums, and the way friends and family view them. Patients who receive genetic testing also need access to genetic counseling, which can make results more useful and less daunting.

Privacy is another concern for patients with rare vision diseases. The Genetic Information and Nondiscrimination Act of 2008 protects one aspect of citizens' genetic privacy. The law prohibits most employers and health insurers from discriminatory use of information as it relates to employment and health coverage.

The legislation, unfortunately, does not entirely solve patients' privacy concerns. Although most employers and insurers are prohibited from accessing genetic information, patients can still be legally discriminated against in life and long-term care insurance.[6] Providers must be sure to inform patients of these facts as they explore genetic testing options.

Coverage can pose another barrier. Genetic testing can be expensive, and insurance companies may not adequately cover the costs for some patients.

Policy Solutions for Genetic Counseling and Education

Shortening the timespan between initial symptoms and definitive diagnosis is key to improving outcomes for patients with rare vision diseases. Genetic testing is one of the best ways to reach a diagnosis faster, but patients are not always aware of this option.

Working with a genetic counselor can help patients understand the potential value of genetic testing and determine whether it's right for them. Genetic counselors can help patients cope with any preliminary anxiety, manage expectations throughout the testing process and course of treatment, and act as the point person to communicate results. In addition to supporting patients as they process the emotions of receiving a genetic diagnosis, genetic counselors can also identify support groups, offer educational resources and connect newly diagnosed patients with fellow patients who face a similar disease.

Genetic counselor shortages could be mitigated by policies that allow counselors to be reimbursed for their services. Members of Congress have introduced bills that would provide coverage for genetic counseling services under Medicare, but these bills have not been passed into law. Patients need their insurers to cover this indispensable service as part of an overall treatment plan.

Simple educational tools such as videos, pamphlets and infographics can also provide baseline information for patients and inform their decision about genetic testing.

Geographic, Socioeconomic and Racial Inequities

Another barrier to diagnosing and treating a rare vision disease may be geography, race or broader socioeconomic factors.

Qualified specialists are often located in major cities, leaving some patients in rural areas with significant transportation challenges. Many patients and their caregivers cannot afford the travel costs or cannot take time off work for appointments and treatments. This can often be an even bigger obstacle for patients who cannot drive because of disease symptoms that impair their vision.

Historically marginalized populations have faced these and other challenges. In fact, a U.S. National Health Interview Survey concluded that disparities in access to genetic testing persist — with Hispanic, uninsured, non-citizen and less educated patients among the most affected.[7] Even if patients live in or near a major city that has medical specialists, they may lack reliable transportation. Being uninsured or underinsured can worsen these problems.

As new and innovative treatments for rare vision diseases emerge, barriers like these may keep new treatments out of reach.

Policy Solutions for Geographic, Socioeconomic and Racial Inequities

Some geographic, socioeconomic and racial inequity challenges can now be alleviated through telehealth, sometimes referred to as telemedicine. Telehealth is the use of electronic information and telecommunication technologies to support long-distance clinical health care, patient and professional health-related education, health administration, and public health. Since the COVID-19 pandemic, telehealth has expanded as a tool to provide medical care remotely.[8]

With telehealth, patients can access specialists from a distance, even from across the country if necessary. There are some limitations, however. Not everyone has broadband access, access to a computer or smartphone, or knows how to use the necessary software.

Non-English support has also been limited in telehealth platforms, which can be isolating for underserved communities.

Insurance coverage for telehealth appointments is also evolving. Some states are changing their licensing laws to allow health care providers to treat patients across state lines. At least 43 states and the District of Columbia have parity laws requiring private health plans to cover telehealth services like virtual visits. As these provisions spread across the country, still more patients with rare vision conditions can benefit from expanded treatment options.[9]

Innovation Barriers

Patients with rare vision conditions generally have few treatment options. Patients may be forced to rely on off-label treatments or go without treatments altogether. That's why innovation is so critical. New treatments for rare diseases often qualify for one of the FDA's accelerated pathways, such as Fast Track, Priority Review or Accelerated Approval. These approaches allow the FDA to expedite the approval process, filling unmet needs by getting innovative treatments to patients more quickly. In the case of the Accelerated Approval Program, the FDA allows for the use of surrogate or intermediate clinical endpoints to evaluate the safety and efficacy of medications for serious conditions with few or no treatment options. Following approval, companies continue conducting clinical trials to confirm the drug's safety and efficacy.

In some cases, however, policymakers are working to limit coverage for medications approved through the FDA's Accelerated Approval Program. Restrictions like these constitute a major setback.

Patients with rare diseases, including rare vision conditions, generally have few treatment options.

They rely on programs like the Accelerated Approval Pathway to gain access to life-changing medications.

The FDA also has authority to grant orphan drug designation to innovative medications that prevent, diagnose and treat a rare disease. Despite the program's success, it is critical to ensure that we protect innovation so patients living without an available treatment can access further innovative treatments.[10]

Policy Solutions for Innovation Barriers

Orphan drug designation can qualify manufacturers for tax credits, market exclusivity or exemption from drug user fees. As a result, hundreds of drugs exist under this designation, mostly for rare diseases.[11] Expanding access to these orphan drugs would create better outcomes for patients with a rare disease. The FDA Accelerated Approval Program must also be protected to expand access to life-changing treatments. Policymakers must work to ensure that patients have access to breakthrough drugs as they're developed.

Insurance Barriers

Sometimes health plans use bureaucratic tactics to reduce insurers' costs — frequently at the expense of patient health. These practices are known as utilization management. Utilization management can be particularly harmful for people with rare vision conditions, given the need for timely and effective treatment.

Prior authorization is one well-known example of utilization management. A doctor may prescribe a treatment, but the patient's insurance may withhold coverage for that treatment until the patient's health care provider submits specific forms and demonstrates that the health plan's criteria have been met. The process can drag on for days or even weeks, especially if the health plan denies the request and the patient's provider has to appeal. In the meantime, the patient goes without treatment.

A related and commonly used utilization management tool is known as step therapy. Patients are required to "fail" one or more treatments, typically those that are less expensive to the insurer, before they can access their physician-prescribed treatment.

Patients who change insurance may be forced by the new insurance company to go through the medically unnecessary step therapy yet again, leading to further delays.

Another health insurance cost-cutting practice is non-medical switching. This occurs when a 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, almost 40% of patients said their new treatment was not as effective as their original. Nearly 60% had complications.[12]

Utilization management tactics are extremely burdensome for patients. It may take years for rare vision patients to receive a formal diagnosis, and then they may have to wait weeks or months to access their prescribed treatment. Utilization management tactics mean more paperwork for the health care provider, potential disease progression, longer delays and more stress for the patient.

Policy Solutions for Insurance Barriers

Many state legislatures have taken action to reduce barriers to care. As of June 2023, 36 states have passed laws to address step therapy and 40 states have passed laws to address prior authorization.

It is imperative that stakeholders continue to monitor and weigh in on utilization management reform by submitting comment letters and testimonies, as well as calling on policymakers to protect patient access.

Policymakers must ensure that incentives for drug manufacturers keep pace with regulatory policy to encourage a steady stream of accessible medications for rare diseases.

Conclusion

Addressing the unmet needs of the rare vision community begins with increasing awareness among patients, caregivers, policy makers and health care providers.

Resolute policymaking is needed to encourage innovation and reduce insurance barriers to treatment. Unmet needs for people with rare vision conditions are multifaceted and widespread, but solutions are within reach.

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The Vision Health Advocacy Coalition promotes patient-centered policies that make services, devices and life-changing treatment accessible for people with vision conditions.

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