Friedreich's Ataxia

A Rare Condition Facing Common Access Challenges

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AfPA | Alliance for Patient Access

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Introduction

Friedreich's ataxia (FA) is a rare progressive neurological condition that usually presents in childhood with balance and gait issues but ultimately results in life-altering fatigue, loss of mobility, and for many, multi-system failure and premature death due to cardiomyopathy.[1]

There are currently over 10,000 rare diseases, 95% of which have no approved treatments.[2] FA is one of the few with a treatment option that has been developed and approved specifically for the disease. This treatment, omaveloxolone, slows disease progression but does not address the underlying genetic cause of FA. Ongoing innovation is needed.

Individuals with FA face many of the challenges that are common for rare diseases, including a long diagnostic odyssey, limited access to care and specialists, and burden and delays in utilization management. All stakeholders, including the patient community, providers, researchers and policymakers, must work together to address these challenges and find ways to expedite getting treatments to patients.

About Friedreich's Ataxia

FA is a rare genetic condition that impacts roughly 15,000 patients globally.[3] The onset of symptoms typically begins between the ages of five and 15, while about 25% of people experience symptom onset in adulthood. The average life expectancy of someone with FA is 35 years.[4]

FA is the result of a genetic mutation that impacts how the body creates a protein called frataxin. The protein supports the mitochondria's creation of energy. Without appropriate levels of frataxin, cells do not have enough energy to function and can become damaged over time. Cells that need a lot of energy to function properly, such as neurons of the brain and spinal cord and cells of the heart muscle, are most affected by the genetic mutation that causes FA.[5]

Symptoms of Friedreich's Ataxia

Individuals with FA can experience various symptoms, including:

Progressive ataxia,
a loss of coordinated movement

Cardiomyopathy,
thickening of
the heart muscle

Cardiac arrhythmia,
abnormal heart rhythm

Scoliosis

Diabetes

Neuropathy

Fatigue

Vision & hearing loss

Dysarthria,
slurring of speech

Dysphagia,
difficulty
swallowing

Mental health conditions,
such as depression or anxiety

Muscle pain
& spasms

Osteoporosis,
low bone density

Pes cavus,
high arch of the foot[6]

These symptoms can place a heavy burden on patients, both physically and mentally. Over time, someone with FA will transition through several mobility devices, from a walker or cane to a manual wheelchair to an electric wheelchair.

As the disease continues to progress, they will begin to need assistance with activities of daily living, including showering, housekeeping, and eating, which often require the employment of personal care assistants.

In the end stages of the disease, people with FA often experience vision and hearing loss and struggle to communicate with their loved ones due to the effect the disease has on their speech. Nearly all people with FA experience cardiac disease including cardiomyopathy, which can lead to heart failure and cardiac arrhythmias. Cardiac disease is the cause of death in most people with FA.

Treating FA requires multiple interventions to minimize health issues and maximize mobility and quality of life. Before an individual can receive appropriate treatments, however, he or she must first receive the correct diagnosis.

The Diagnostic Odyssey

Someone with FA may experience an extensive journey to diagnosis, a difficult process known as "the diagnostic odyssey." Rare disease patients wait an average of six years or more for an accurate diagnosis, particularly because rare conditions like FA often require specialized knowledge and testing.

Patients must be referred to the right specialist, who must order the appropriate tests to achieve the right diagnosis. But along the way, patients may encounter:

- Specialists who are unfamiliar with the relevant symptoms for FA, resulting in the patient getting stuck in a loop of referrals;
- Specialists who do not carefully listen to the patient and makes incorrect referrals;
- Long and costly travel to specialists;
- Lack of access to genetic testing and genetic counselors;
- And misdiagnosis.

Misdiagnosis not only causes emotional distress but also delays the individual from accessing experts knowledgeable in FA who can best manage symptoms and connect them to relevant treatments and clinical trials.

Common misdiagnoses for FA are multiple sclerosis or a neurological disease called Charcot Marie Tooth.

The diagnostic odyssey is also exacerbated by the fact that FA symptoms typically begin in childhood. The first point of contact at the onset of symptoms is usually a pediatrician. A general pediatrician may not be familiar with the signs and symptoms of FA and may mistake initial symptoms for normal variations in childhood motor development.

The diagnostic odyssey for FA has decreased in recent years thanks to genetic testing, with individuals under 10 years old in the United States receiving a diagnosis within two years of symptom onset. But many patients, particularly those with adult onset of symptoms and patients who live outside of the United States, still wait an average of eight years or more to reach the correct diagnosis.

Genetic testing and counseling are important steps in shortening the diagnostic odyssey for individuals with FA. Policies that support access to genetic testing and counseling are imperative to expedite the FA diagnosis and life-altering treatment.

Treatment Challenges

Once an individual has an FA diagnosis, the next step is to seek appropriate treatment. Getting access to the right treatment, however, can be a complicated path.

For a long time, there weren't any FDA-approved treatments for FA patients. Treatment instead focused on disease and symptom management.

This changed in 2023, when the first treatment for FA, omaveloxolone, was approved by the FDA. Omaveloxolone slows the progression of neurological symptoms, allowing individuals to retain more motor function for longer.

Although omaveloxolone is not a cure for FA, it is an exciting step forward for the FA community. Omaveloxolone alone is not enough, however.

FA requires comprehensive treatment. Mobility aids, medication to manage cardiac symptoms and neuropathy, and scoliosis surgery are examples of the kinds of clinical management FA requires.

> Omaveloxolone is an **exciting step forward** for the FA community.

Hurdles To Treatment and Solutions

Despite the fact that omaveloxolone has been approved by the FDA, and experts have created clinical care guidelines to help providers treat FA, patients and providers may struggle to access appropriate treatment for many reasons.

Clinician Availability & Knowledge Gaps

Finding available FA clinicians is one of the greatest challenges that FA patients experience. Few general providers fully understand FA and can correctly direct someone for diagnosis and treatment. There are also few FA specialists, and getting connected to the right specialist may take a significant amount of time.

Increased awareness will benefit patients and health care providers alike. Recognizing the signs and symptoms can shorten the diagnostic odyssey and expedite treatments options. It could also reduce associated mental health stressors and connect the individual to other people with FA who understand the difficulties of the disease. Additionally, health care providers could be exposed to new tools, treatments and devices that can help their patients.

All stakeholders can advance this effort. The FA patient community is the expert in this disease and educates both clinicians and policymakers by sharing their lived experiences. Clinicians and policymakers, armed with this insight, can facilitate changes that reduce the access burden.

Limits on Innovation

Innovation led to the first approved treatment for FA. Further innovation will continue to improve disease management and treatment. Policies that support robust and timely funding of federal research agencies, stronger collaboration between drug developers and regulators, and better visibility throughout coverage decisions would all incentivize new treatments being developed.

Limitations on Clinical Trials

Proper design of clinical trials is also crucial to bringing new treatments for FA forward. Since FA is a progressive condition, treatments are most beneficial when delivered as early as possible. Due to safety concerns, children are often excluded from clinical trials until the drug has been tested in adult populations. This paradigm does not work for rare pediatric disorders like FA, where without intervention, children face worsening symptoms and disease progression. Involving children with FA in clinical trials is imperative for the approval of treatments for those children.

Regulations allowing children with rare or chronic conditions to participate in early-stage clinical trials need to be supported.

Transportation & Telehealth Challenges

FA specialists are few and far between. They are spread across the country and meeting with them may be difficult for patients. There is the loss of work, school and family time; the travel and lodging costs; the need for caregiving services; and the reality that mobility devices are often broken by airlines.

All these hurdles present serious barriers in accessing appropriate clinical care and may delay genetic testing, medical procedures and treatments for FA.

Telehealth is a valuable tool that can help many with FA. It can bridge the distance between the individual and the health care provider. While some appointments and tests must be in person, many can be done without the immense burden of travel to and from a specialist.

Telehealth visits, however, are not always covered under insurance and are often excluded by Medicaid. A specialist in a neighboring state may be the best option but could be cost prohibitive without coverage. An additional bonus of covering telemedicine is that it allows a FA specialist to work with the person's care team at home. Telehealth is an important aspect of health care that deserves support from all stakeholders.

Mental Health Challenges

FA is a devastatingly relentless disease that often leads to mental health challenges for the individual and caregivers. Research demonstrates that depression is common in FA. The mental and emotional toll on parents and loved ones who care for individuals affected by rare and progressive conditions like FA is well documented.

Factors that affect the mental health of those impacted include the burden of long diagnostic odysseys, an uncertain future in the face of continuously progressing symptoms, and isolation that comes from having a rare disease that limits mobility, energy and sometimes speech, vision and hearing.

Mental health challenges in FA are multifaceted, but taking steps to address barriers to diagnosis, funding research for future FA treatments and raising awareness of FA are sure to improve mental health outcomes for all.

Utilization Management

While an approved treatment is available for FA, individuals and their providers can face difficulties in getting coverage for it. Health care benefits are often monitored by "utilization management," which is "a set of techniques used by or on behalf of purchasers of health care benefits to manage health care costs by influencing patient care decision-making through case-by-case assessments of the appropriateness of care prior to its provision."[7]

When access to medication is limited by roadblocks such as prior authorization, step therapy and non-medical switching, it creates another burden for patients by delaying necessary care. Furthermore, the added administrative steps place an unnecessary burden on patients and providers.

Stakeholders can work together to restrict utilization management tactics and help patients access treatments more quickly. Policies that limit the use of prior authorization request and programs that ensure access to affordable medication are crucial. Individuals who are already dealing daily with such a devastating condition should not have to also struggle to access the only FDA-approved FA treatment that could help them.

Pharmacy Benefit Managers

Another coverage challenge is pharmacy benefit managers (PBMs). PBMs work with health plans to determine which medications are covered by the insurer. Their focus is to contain costs for insurers, and they accomplish that in several ways. PBMs develop lists of approved drugs, or formularies. When a drug is excluded from the list, it is not covered by that insurance and patients are often unable to afford or access the medication. New treatments for rare diseases like FA are often excluded by PBMs. PBMs can also establish copay and coinsurance plans that transfer some of the financial cost to the patient.

If a PBM refuses to cover a medication, or establishes an unfair copay plan, there is no recourse for the FA community. This process is ripe for legislative intervention.

Conclusion

FA is an incredibly difficult diagnosis to receive. As the lived experiences of the FA community show, diagnosis is often followed by common health care challenges including access to appropriate care and necessary treatments.

All stakeholders, including the patient community, providers, researchers, or policymakers have a responsibility to advance policies and procedures to minimize those access hurdles.

The power of change calls for all parties to increase awareness of conditions like FA, encourage innovation for new treatments, help reduce diagnostic and access barriers, and support individuals to achieve the improved quality of life that medical intervention can provide.

Resources

1. What is Friedreich's ataxia? Friedreich's Ataxia Research Alliance. (2024, July 19). https://www.curefa.org/understanding-fa/what-is-friedreichs-ataxia/.
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AfPA | Alliance for Patient Access

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

FARA | Friedreich's Ataxia Research Alliance

ABOUT THE FRIEDREICH'S ATAXIA RESEARCH ALLIANCE (FARA)

The Friedreich's Ataxia Research Alliance (FARA) is a national, public, 501(c)(3), non-profit, organization dedicated to the pursuit of scientific research leading to treatments and a cure for Friedreich's ataxia.

CureFA.org