UNMET NEEDS

in hATTR Amyloidosis in Europe

EAfPA
European Alliance for Patient Access

Introduction

People living with rare diseases face a unique set of challenges, from reaching a timely and accurate diagnosis to accessing treatment. Although individual rare diseases may impact only a small population, there about 30 million people living in Europe with one of 6,000 identified rare diseases.[1]

One such rare disease is hereditary transthyretin (hATTR) amyloidosis. It is caused by a hereditary gene mutation that leads to a build-up of abnormal protein in different bodily organs. The disease causes debilitating cardiovascular, neurological, renal or other symptoms. As the disease progresses, patients can experience difficulty walking, fatigue, chest pain, and weakness and pain in their limbs, making it difficult to continue with their normal lives.

Due to the complex nature of hATTR amyloidosis, and the impact on different organs, reaching a diagnosis can be a complex process. When hATTR amyloidosis is diagnosed and treated in a timely manner, however, disease progression can be stopped, giving patients hope for a better quality of life.

Yet gaps in access and unmet needs keep patients from timely treatment. This white paper outlines five unmet needs that advocates agree are inhibiting optimal care for hATTR amyloidosis patients in Europe:

- **Early & Accurate Diagnosis**
- **Equitable Access to Treatment**
- **Guidance on Genetic Testing**
- **Support for Families & Carers**
- **Broad Access to Guidelines**

Unmet Needs

Early & Accurate Diagnosis

An early, accurate diagnosis for hATTR amyloidosis is often difficult to attain due to the complexity and wide array of symptoms, as well as the lack of awareness about the disease.

For most patients, the first point of care is their primary care physician. Unlike a rare disease expert, however, most primary care physicians are not trained or familiar enough with symptoms to identify rare diseases. Patients are often misdiagnosed.

Even when a patient seeks treatment from a specialist, that physician may investigate only the symptoms that fall within their specialty. While treating the symptoms in isolation, specialists may struggle to recognise the underlying disease, ending in a misdiagnosis.

As a result, hATTR amyloidosis patients often experience a lengthy diagnosis process, seeing five or more clinicians before reaching an accurate diagnosis.[2]

With a degenerative disease like hATTR amyloidosis, time is of the essence. Lengthy periods during which patients are misdiagnosed or go undiagnosed simply delay the onset of treatment. The delay in diagnosis can also intensify patients' anxiety.

Being able to put a name to one's symptoms often brings some relief, even when the diagnosis is for a rare disease as serious as hATTR amyloidosis.

> **"Having treatments changes everything. You want to know the diagnosis as early as possible to allow the patients to access treatment."**
>
> **Ashutosh Wechalekar, MD,** ***Haemotologist***

Steps Forward

There are ways to address delays and misdiagnoses. Educating non-specialist nurses and physicians, those who see and treat patients regularly, can be a good place to start. Teaching clinicians about some of the "red flag" symptoms can also help to keep the potential for amyloidosis top of mind.

With a hereditary disease like hATTR amyloidosis, it is also important that patients and their clinicians are aware of family history. This can help to reduce the time required to reach a diagnosis.

Equitable Access to Treatment

Though treatments for hATTR amyloidosis exist, patients may struggle to access them.

Currently in Europe, there are three different treatments approved by the European Medicines Agency to treat hATTR amyloidosis. Two of these medications are approved to treat the polyneuropathy symptoms of hATTR amyloidosis and one is approved to treat the cardiomyopathy symptoms. These are welcome options because historically many amyloidosis patients have had to rely on transplants as the standard of care due to lack of available treatments. Although the treatments are not curative, they can slow disease progression and drastically improve quality of life for patients.

> **"When the opportunity comes to be part of the health technology assessment process, we must get in there and contribute. It's important that we put across the human side."**
>
> **David Gregory,**
> ***UK ATTR Amyloidosis' Patients Association***

EMA approval, however, does not necessarily equate to access for all patients. Many patients across Europe still struggle to access these treatments depending on their country of residence. Much like treatments for other rare diseases, those for amyloidosis can often be expensive. Patients may find that, although the treatment has been approved by the EMA, their government or health system has deemed that the treatment is not cost effective, making access difficult or impossible.

For these treatments to provide optimal benefit to patients and to deliver maximum value to health care systems, patients need timely access.

Steps Forward

When looking to address inequitable access to treatment, patients and advocacy organisations have an integral role to play. Patient organisations need to ensure that they are a part of this decision-making process, letting policymakers understand the impact that these treatments could have on the lives of people living with the disease.

Guidance on Genetic Testing

Genetic testing can be an important tool for identifying and managing a hereditary disease like hATTR amyloidosis, but patients and their families often find little support in this area.

Genetic testing allows family members to test for the inherited mutation that causes hATTR amyloidosis and can empower them to seek treatment early. However, genetic testing can also have implications for factors such as the cost of health insurance and access to life insurance coverage.

Weighing up the potential benefits and drawbacks of genetic testing is challenging, and patients and their families need guidance throughout the process. Instead, patients and family members are often left to navigate these decisions on their own.

People considering genetic testing need to be fully informed of all potential outcomes and given the necessary time to make the decision that is best for them and their family.

Steps Forward

With the recent development of treatments for hATTR amyloidosis, the need for early diagnosis is as crucial as ever. And the role that genetic testing can play has greatly increased. Raising awareness about the option for genetic testing can help patients reach a diagnosis and access treatment at an earlier stage. It remains important, however, that patients and their families receive the necessary support prior to, during and after the genetic testing process.

“ It is important for the clinician to always ask the question ‘Has anybody else in your family had these signs or symptoms?’ ”

Tootie Bueser, RN
British Association for Nursing in Cardiovascular Care

Support for Families & Carers

Amyloidosis brings a heavy burden on families and carers, though their role is too often overlooked.

Patients with hATTR amyloidosis often have difficulty walking and experience swollen ankles and limbs, weakness and fatigue. These symptoms make it difficult to live independently. The severe burden of disease impacts not only the life of the patient but also the lives of their family members and carers. Caregivers may need to quit or change their jobs to devote more time to caring for their loved one with hATTR amyloidosis. Adult children may need to move home to care for their parents. Patients who were once independent and active may no longer be able to do the basic tasks of caring for themselves.

Family and carers should be acknowledged as a core part of the patient's care team and integrated into the treatment process. They also deserve adequate educational and emotional support, including a place where their questions can be answered and access to someone who can provide them with much-needed emotional support.

> **"Patients and carers need a local point of support – especially in the early days of diagnosis when their world has been turned upside down."**
>
> **Rosaline Callaghan,**
> *Amyloidosis Ireland Support Group Patients Association*

Steps Forward

Ensuring that families and carers, along with patients, have a local and accessible system of support is important. Having someone to walk them through the process and answer initial questions can be especially valuable. One important step forward is making sure that speciality centres and hospitals are aware of the different support groups in the region. This way, when patients and their families leave the hospital, they have somewhere to turn.

Broad Access to Guidelines

Though guidelines for the diagnosis and treatment of hATTR amyloidosis have been developed, they are rarely accessible to people outside the health care system.

> **"A valuable piece of work for advocates would be to take the guidelines and work with the experts to create something that's easy to read and understand."**
>
> **Carys Barton, RN**
> *British Society for Heart Failure Nurse Forum*

The guidelines remain largely limited to academic and scientific spheres. As a result, advocacy groups and lay people don't always know that the guidelines exist or where to find them.

Advocacy groups can serve as helpful conduits, passing important information like these guidelines to patients. The academic and scientific community that develops the guidelines should expand the reach of these guidelines – and should use advocates to do so. Broader availability will help to improve awareness about hATTR amyloidosis and could spur necessary policy change.

Steps Forward

Guidelines can often be difficult to understand, even for the clinicians that specialise in the area. Providing summarised or revised versions of the guidelines and highlighting the key messages can make them more accessible and easier to understand across a wider population.

Conclusion

Patients with hATTR amyloidosis face many major hurdles. They often spend years visiting different specialists just to put a name to their disease. They can then struggle to gain access to treatment, even with three medications approved on the European level. Meanwhile, they may wrestle alongside family members with questions of genetic testing, often with little guidance.

For policymakers and advocates to facilitate better outcomes for patients, they must address several factors.

Those factors include:

- The need for early and accurate diagnosis
- Inequitable access to treatment
- Lack of guidance and support on genetic testing
- Support for family and carers
- Limited awareness and application of guidelines

Growing awareness and new treatment options offer hope for hATTR amyloidosis patients and families across Europe. **Addressing unmet needs can go a long way towards translating that hope into better treatment outcomes and improved quality of life.**

References

1. *Eurordis Rare Diseases Europe. What is a rare disease? 2020. Retrieved from: https://www.eurordis.org/content/what-rare-disease*
2. *Lousada, I, Maurer M S, Warner M, Guthrie S, Hsu K, Grogan M. Amyloidosis Research Consortium Cardiac Amyloidosis Survey: Results from Patients with AL and ATTR Amyloidosis and Their Caregivers. American Research Consortium. 2019. Retrieved from: https://arci.org/wp-content/uploads/2019/09/FINAL-2019-HFSA-Cardiac-Amyloid-Poster-2019.09.10-1.pdf*

Amyloidosis Working Group

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