Transthyretin Familial Amyloid Polyneuropathy:
A Debilitating Disease Affecting Patients and Caregivers

Transthyretin Familial Amyloid Polyneuropathy: Do You Have a Family Connection?

Transthyretin familial amyloid polyneuropathy (TTR-FAP) is a rare, genetic, and often fatal neurodegenerative disease estimated to affect 10,000 people worldwide.\(^1\)

**If left untreated, TTR-FAP causes death due to complications from the disease within 10 years of the onset of symptoms, on average.\(^2,3\)**

TTR-FAP is inherited from a biological parent who may or may not have the disease but carries the genetic mutation.\(^4,5\)

Since TTR-FAP is genetically inherited, individuals with a family history of the disease should be aware of the symptoms and work with their doctor so that, if symptoms occur, a diagnosis can be made as early as possible. Genetic testing can play a critical role in early diagnosis.\(^4,5\)

The Impact of TTR-FAP on Caregivers

Living with TTR-FAP is physically, emotionally, and financially challenging, not only for patients but also for their caregivers.\(^6\)

TTR-FAP is irreversible and progressive\(^1\). It can be frightening for family members to watch parents, grandparents, or other family members suffer from this debilitating disease.

As a caregiver, it is important to maintain your own health. This means understanding the symptoms of TTR-FAP and speaking with your doctor about your risk.

In a survey of patients and caregivers, a majority of caregivers (61\%) reported having transthyretin amyloidosis.\(^6\)

Non-Specific Symptoms Make Diagnosis Challenging

TTR-FAP symptoms are non-specific and often start with numbness in the feet, a decrease in temperature sensation, and pin-prick feelings—symptoms that can also be caused by injury, infection, other neurological conditions, or even more common diseases, like diabetes.\(^7\)

This, plus low awareness around the disease, makes diagnosis challenging. It can take several years from when symptoms start to receive an accurate diagnosis.\(^1,2,5,8,9\)

Since the disease is progressive, early diagnosis and treatment—before symptoms progress and become severe—are important.\(^10,11\)

People in the late stage of TTR-FAP often lose the ability to walk, eventually becoming bedridden and unable to care for themselves.\(^4,12\)
**What to Watch For**

There are clusters of clinical symptoms that should raise suspicion of a possible TTR-FAP diagnosis. These “red flag” symptom combinations include the presence of progressive peripheral sensory-motor polyneuropathy and at least one of the following:

- Family history of neuropathy
- Early dysfunction of the autonomic nervous system, which regulates unconscious bodily functions such as breathing, heart rate, and digestion
- Cardiac involvement
- Diarrhea
- Constipation
- Unexplained weight loss
- Carpal tunnel syndrome
- Renal impairment *(also known as kidney failure or renal insufficiency)*
- Vitreous opacity *(“floaters” in the eye)*

If TTR-FAP is suspected, genetic testing may be performed.13

**Talk to Your Doctor**

It is critical for you to find the right support to help you manage the impact of the disease as well as the potential added burden of feeling scared, hopeless, or insecure.

Building a strong dialogue with your doctor and making each visit productive and informative can help. Before each medical visit, make sure you:

- Track symptoms you are experiencing, along with their frequency and severity
- Prepare a list of questions and details of your family history
- Use the TTR-FAP Dialogue Builder at [www.ttr-fapconnection.com/dialogue-builder](http://www.ttr-fapconnection.com/dialogue-builder) to help guide your conversation

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