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. While TTR-FAP has been estimated to affect 10,000 people worldwide, the actual prevalence may be much higher.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 Since TTR-FAP runs in families, 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.

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

If You Are a Caregiver: The Importance of Taking Care of Yourself

Did you know that a majority of caregivers (61%) have reported having symptoms of transthyretin amyloidosis?6 We understand that the prospect of possibly having TTR-FAP, especially when you care for a family member with the disease, may be overwhelming and frightening.

However, it’s important to take time for yourself and maintain your own health. This includes educating yourself about the symptoms of TTR-FAP and talking to your doctor about your risk.

TTR-FAP is a relentlessly progressive disease,6 so the earlier you speak with your doctor and seek treatment, the better — before symptoms progress and become severe.11,12

Nonspecific Symptoms Make Diagnosis Challenging

TTR-FAP symptoms are nonspecific 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.2, 5, 8, 9, 10
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)¹³

Talk to Your Doctor

If you have a family history of TTR-FAP, talk to your doctor about your risk. If suspected, the diagnosis of TTR-FAP can be confirmed by genetic screening, which is a simple blood test.⁴,⁵

It is important to build a strong dialogue with your doctor and make each visit a productive and informative one. Before each visit, make sure you

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

Visit www.ttr-fapconnection.com for more information and additional resources.