Recognizing Transthyretin Familial Amyloid Polyneuropathy: Connecting the Dots to Increase Awareness

What is Transthyretin Familial Amyloid Polyneuropathy?

Transthyretin familial amyloid polyneuropathy, or TTR-FAP, is a genetic neurologic disease. It is also relentlessly progressive, so early diagnosis and treatment are important to managing this disease.¹

TTR-FAP is caused by a mutation in the transthyretin (TTR) gene, which can result in abnormal and unstable TTR proteins. These abnormal proteins can then build up and form structures called amyloid. Amyloid may deposit in a specific part of the nervous system called the peripheral nervous system, which can lead to a decline in neurologic function. Amyloid may also deposit in the heart, kidney, eyes, and gastrointestinal tract, leading to malfunction in these areas as well.² ³

A Look at the Numbers

Estimated to affect 10,000 people worldwide,⁴ although prevalence may be much higher. A person has a 50% chance of inheriting TTR-FAP if a biological parent has it⁵. Disease onset may occur as early as age 30.¹ If left untreated, TTR-FAP causes death due to complications from the disease within 10 years. On average, it takes 4 years from when symptoms start to receiving an accurate diagnosis.¹ ² ⁶ ⁸ From the onset of symptoms, on average⁶ ⁸

Who is Affected by TTR-FAP?

TTR-FAP is found worldwide and has been reported in 36 countries, including Portugal, Brazil, Japan, Sweden, the United States, Taiwan, France, Italy, Spain, and Germany.⁴ TTR-FAP affects men and women equally.¹⁰
What are the Symptoms?

At the onset of disease, people with TTR-FAP experience neurologic changes, including:

- numbness in the feet
- decreased temperature sensation
- pin-prick feelings

TTR-FAP also affects the autonomic nervous system, which controls involuntary processes in the body. These symptoms may include:

- dizziness or fainting
- bouts of constipation that alternate with diarrhea
- abnormal heartbeat
- unintentional weight loss

Early diagnosis and treatment—before symptoms become severe—are critical to delaying disease progression and maintaining quality of life.12,13

TTR-FAP worsens over time and can progress rapidly. Without treatment, people with TTR-FAP may lose the ability to walk, and eventually become bedridden. These progressive neurologic symptoms are referred to as peripheral sensory-motor polyneuropathy.14,15

How is the Disease Diagnosed?

TTR-FAP may be challenging to diagnose. 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.16

People with a family history of the disease should talk with their doctors about their risks. TTR-FAP is inherited from a parent with the genetic mutation, even if this parent never showed signs or symptoms of the disease. TTR-FAP may also skip 1 or more generations.5,7

If suspected, the diagnosis of TTR-FAP can be confirmed by genetic screening, which is a simple blood test to identify the presence of mutations in the TTR gene followed by tissue biopsy to determine the presence of amyloid deposits.5,17

Learn more: visit www.ttr-fapconnection.com for helpful resources including:

A “Dialogue Builder” to prepare for discussions with health care professionals

Answers to common questions

A list of helpful groups and organizations