

## FACT:

# Identifying Early Signs of Friedrich Ataxia Can Help Speed Time to a Definitive Diagnosis

Early symptoms of Friedrich ataxia (FA), typically appearing between the ages of 10 and 15, can overlap with other conditions.<sup>1,2</sup> **FA is the most common inherited ataxia**,<sup>3</sup> making it essential to think FA **FIRST** when you see any combination of these signs and symptoms.



### Falls<sup>1</sup>

(gait ataxia)



### Imbalance<sup>1,4</sup>

(proprioception loss)



### Reflex loss<sup>4</sup>

(areflexia)



### Sensation loss<sup>4</sup>

(peripheral neuropathy)



### Tiredness<sup>3</sup>

(chronic fatigue)

## A Genetic Test That Includes a Friedrich Ataxia Repeat Expansion Analysis Confirms an FA Diagnosis<sup>5</sup>

Nearly **98%** of FA cases are caused by a genetic variant within the frataxin (FXN) gene called a GAA triplet-repeat expansion, with about 2% attributable to point mutations.<sup>4</sup>

Not all reference laboratories offer a genetic test that can detect these expansions, so ordering the right test is crucial.

Confirming a diagnosis accelerates your patients' connection to multidisciplinary supportive care and empowers them to make informed decisions.



Get additional information and resources at [ThinkFA.com](https://www.thinkfa.com).



# FACT:

## Friedreich Ataxia (FA) Can Be Overlooked Because Patients Often Describe Vague Initial Symptoms<sup>1,6</sup>

### WHAT PARENTS OF YOUNGER PATIENTS MAY SAY:

"I notice that she often falls down."<sup>1,3</sup>

### WHAT OLDER PATIENTS WITH LATER-ONSET FA MAY SAY:

"People say I look intoxicated when I walk."<sup>1,3</sup>

### Is it clumsiness or is it **gait ataxia**?<sup>4</sup>

"At night, he struggles walking up the stairs."<sup>7</sup>

"If I'm in a dark room, I feel like I'm on a rocking boat."<sup>3</sup>

### Is it a passing difficulty or is it **loss of proprioception**?<sup>4</sup>

"Her handwriting is getting more difficult to read."<sup>1</sup>

"I can't type text messages lately."<sup>1</sup>

### Is it a temporary challenge or is it **loss of fine motor skills**?<sup>1</sup>



Recognizing early signs of FA can help you diagnose sooner and potentially make a big difference in a patient's life.

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**References:** **1.** Parkinson MH, Boesch S, Nachbauer W, Mariotti C, Giunti P. Clinical features of Friedreich's ataxia: classical and atypical phenotypes. *J Neurochem*. 2013;126(suppl 1):103-117. **2.** Wallace SE, Bird TD. Molecular genetic testing for hereditary ataxia: what every neurologist should know. *Neural Clin Pract*. 2018;8(1):27-32. **3.** National Institute of Neurological Disorders and Stroke. Friedreich Ataxia Fact Sheet. Updated November 15, 2021. Accessed March 16, 2022. <https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Friedreichs-Ataxia-Fact-Sheet>. **4.** Fogel BL, Perlman S. Clinical features and molecular genetics of autosomal recessive cerebellar ataxias. *Lancet Neurol*. 2007;6(3):245-257. **5.** Schulz JB, Boesch S, Bürk K, et al. Diagnosis and treatment of Friedreich ataxia: a European perspective. *Nat Rev Neurol*. 2009;5(4):222-234. **6.** de Silva RN, Vallortigara J, Greenfield J, Hunt B, Giunti P, Hadjivassiliou M. Diagnosis and management of progressive ataxia in adults. *Pract Neurol*. 2019;19(3):196-207. **7.** Han J, Waddington G, Adams R, Anson J, Liu Y. Assessing proprioception: a critical review of methods. *J Sport Health Sci*. 2016;5(1):80-90.