

FACT:

Identifying Early Signs of Friedreich's Ataxia Can Help Speed Time to a Definitive Diagnosis

Early symptoms of Friedreich's ataxia (FA), typically appearing between the ages of 10 and 15, can overlap with other conditions.^{1,2} **FA is the most common inherited ataxia,³** making it essential to think FA **FIRST** when you see any combination of these signs and symptoms.



Falls¹

(gait ataxia)



Imbalance^{1,4}

(proprioception loss)



Reflex loss⁴

(areflexia)



Sensation loss⁴

(peripheral neuropathy)



Tiredness³

(chronic fatigue)

A Genetic Test That Includes a Friedreich's Ataxia Repeat Expansion Analysis Confirms an FA Diagnosis⁵

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.⁴

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 that can help them maintain independence longer.



Get additional information and resources at ThinkFA.com.

FACT:

Friedreich's Ataxia (FA) Can Be Overlooked Because Patients Often Describe Vague Initial Symptoms^{1,6}

WHAT PARENTS OF YOUNGER PATIENTS MAY SAY:	WHAT OLDER PATIENTS WITH LATER-ONSET FA MAY SAY:
"I notice that she often falls down." ^{1,3}	"People say I look intoxicated when I walk." ^{1,3}
Is it clumsiness or is it gait ataxia?⁴	
"At night, he struggles walking up the stairs." ⁷	"If I'm in a dark room, I feel like I'm on a rocking boat." ³
Is it a passing difficulty or is it loss of proprioception?⁴	
"Her handwriting is getting more difficult to read." ¹	"I can't type text messages lately." ¹
Is it a temporary challenge or is it loss of fine motor skills?¹	



Recognizing early signs of FA can help you initiate intervention 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. *Neuro 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, Burk 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.