

## FACT:

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

Early symptoms of Friedrich's 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)



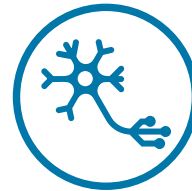
### Imbalance<sup>1,4</sup>

(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's 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 that can help them maintain independence longer.



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

# FACT:

## Friedreich's 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 initiate intervention sooner and potentially make a big difference in a patient's life.

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

**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.

## FACT:

# The mFARS Neurological Exam Provides a Detailed Evaluation of a Patient's Status<sup>1</sup>

**Friedreich's ataxia (FA)** is the most common inherited ataxia.<sup>2</sup> While progression varies by patient, several studies estimate that between 55% and 78% of patients with FA will require a wheelchair within 10 to 15 years after disease onset. For those with more severe FA, this can occur in as few as 3 years.<sup>3</sup>

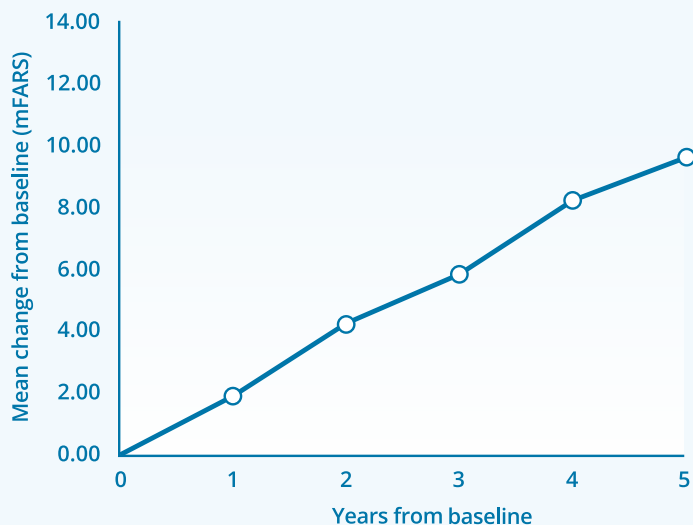
The modified Friedreich's Ataxia Rating Scale (mFARS) is a clinically validated set of assessments that measures FA progression and its impact on a patient's ability to perform activities of daily living.<sup>1</sup> Changes in key mFARS scores help predict time to loss of ambulation and inform management strategy.<sup>4,5</sup>

## The mFARS Assesses Current Status and Helps Predict Future Decline<sup>1,4</sup>

**4:** The number of mFARS evaluation items. Each item is scored to determine the current severity of disability in a specific area<sup>1</sup>

**93:** The maximum mFARS composite score across all 4 items<sup>1</sup>

**65:** Average mFARS score where loss of ambulation occurs<sup>6</sup>



### Patient mFARS Composite Scores Will Typically Increase ~2 Points per Year

Adapted from: Patel M, et al. *Ann Clin Transl Neurol.* 2016;3(9):684-694. In a multicenter natural history study, 812 diagnosed patients were evaluated annually using multiple tests, including FARS and mFARS. Mean length of GAA triple repeat was 636 and mean age of onset was 13.7 years. Results shown are average annual change from baseline in mFARS composite score for overall cohort.

## The mFARS Is Typically Accepted as a Clinical Trial Endpoint

Because of their correlation with disease progression, changes in mFARS scores over time are typically accepted as an endpoint in clinical trials for potential new FA treatments.<sup>1</sup>

A patient's disease severity and rate of progression directly relate to the extent of the genetic triplet-repeat expansion that causes FA. A larger expansion is associated with more serious symptoms and a more rapid decline in function.<sup>3,7</sup>



# The 4 Components of mFARS

The mFARS includes traditional elements of a neuromuscular assessment that specifically focus on a patient's disabilities.

Assessment	Related Daily Activities	Assessment Goals	What This Means for Patients <sup>1</sup>
<b>Bulbar Function</b>	Swallowing or speaking	Measure strength and volume of coughing and clarity of speech. <sup>1,3,7</sup>	Asking the patient to cough 3 times can reveal difficulties with swallowing, catching their breath, or clearing their airways. <b>Clinical/functional impact:</b> Risk of respiratory infection. This component also asks the patient to say 2 phrases, such as: "The traffic is heavy today." and "The president lives in the White House." <b>Clinical/functional impact:</b> Ability to communicate clearly, a key element in maintaining independence.
<b>Upper Limb Coordination</b>	Brushing teeth, typing, pointing, reaching, or turning a doorknob	Measure motor abilities related to tremors, fine motor coordination, and steadiness of hands and arms. <sup>8,9</sup>	The patient performs finger-to-finger, nose-to-finger, finger-chase, and rapid hand movements. <b>Clinical/functional impact:</b> Ability to complete many activities of daily living, including getting dressed and eating, as well as written or electronic communication abilities. These tests can also reveal the presence of tremors.
<b>Lower Limb Coordination</b>	Putting on socks and shoes	Measure coordination of legs and feet. <sup>4</sup>	The patient performs a heel-shin slide and heel-shin tap. <b>Clinical/functional impact:</b> Likely rate of disease progression, which is also closely correlated with upright stability results.
<b>Upright Stability</b>	Walking, sitting in a car, standing in line, or showering	Measure loss of ambulation and impaired ability to coordinate voluntary movements (ataxia). <sup>4,10</sup>	The rate of progression is most evident in this component set. Six components evaluate the amount of time a patient can stand or walk in a steady position without listing or needing assistive devices. Three sitting components assess the patient's seated posture. <b>Clinical/functional impact:</b> Predicts time to loss of function.



Get tools, information, and resources to help your patients at [ThinkFA.com](https://www.thinkfa.com).

**References:** 1. Rummey C, Corben LA, Delatycki MB, et al. Psychometric properties of the Friedreich Ataxia Rating Scale. *Neural Genet.* 2019;5(6):371. 2. 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>. 3. 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. 4. Rummey C, Farmer JM, Lynch DR. Predictors of loss of ambulation in Friedreich's ataxia. *EClinicalMedicine.* 2020;18:100213. 5. Pandolfo M. Neurologic outcomes in Friedreich ataxia: Study of a single-site cohort. *Neural Genet.* 2020;6(3):e415. 6. Patel M, Isaacs CJ, Seyer L, et al. Progression of Friedreich ataxia: quantitative characterization over 5 years. *Ann Clin Transl Neurol.* 2016;3(9):684-694. 7. 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. 8. Johns Hopkins Medical. Neurological Exam. Accessed March 16, 2020. <https://www.hopkinsmedicine.org/health/conditions-and-diseases/neurological-exam>. 9. 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. 10. Friedman LS, Farmer JM, Perlmans S, et al. Measuring the rate of progression in Friedreich ataxia: implications for clinical trial design. *Mov Disord.* 2010;25(4):426-432.

# Friedreich's Ataxia Backgrounder

and Important Considerations

Information that may help you  
manage your patients with FA

# WHAT IS FRIEDREICH'S ATAXIA?

**Friedreich's ataxia (FA)** is an autosomal-recessive, neurodegenerative disease that primarily affects the nervous system but can have multisystemic effects, including cardiomyopathy. People with this disease develop impaired muscle coordination (ataxia) that worsens over time. Though rare, FA is the most common inherited ataxia.<sup>1</sup>

Most patients begin to experience symptoms around puberty (10 to 15 years of age). While presentation of the disease is different in every patient, gait ataxia and general clumsiness are the most common presenting symptoms.<sup>2</sup>

Because of the serious consequences of the disease, patients should be diagnosed as early as possible. An early diagnosis can help manage the symptoms of FA and allow for the building of a multidisciplinary care team tailored to a patient's individual needs.

## The latest research in FA

**The Collaborative Clinical Research Network in Friedreich's Ataxia (CCRN in FA)** is an international network of clinical research centers that work together to allow patients with FA to enroll and participate in research studies and to receive clinical care. While not necessarily involved in day-to-day patient care, CCRN can be a critical resource for patients and neurologists alike. FA specialists at the CCRN have experience working with hundreds of patients with FA, can provide updates on the latest FA research, and can share information on therapies currently in development.

To learn more, visit [CureFA.org/network](https://CureFA.org/network).



## A Closer Look at Common Symptoms of FA

When ataxia is discussed, often a lack of muscle coordination comes to mind. However, it is important to note that FA can also manifest in other ways. Some of the main symptoms observed in patients with FA are outlined below.



### Reflexes, muscle tone, and sensory loss<sup>2</sup>

- Loss of reflexes, particularly in the lower limbs, and extensor plantar reactions are present in almost all patients
- Muscle tone is typically normal or reduced, particularly in the early stages of the disease
- Spasticity occurs in some patients, particularly in the lower limbs. If left untreated, it can cause pain, discomfort, positioning problems, and contractures
- Distal sensory loss is a universal symptom, with most patients losing the ability to sense vibration and joint position



### Gait and limb ataxia<sup>2</sup>

- When limb ataxia is present (an early feature of the disease), daily activities that require fine dexterity become much harder for patients. This causes difficulty with handwriting, washing, dressing, and use of cutlery
- As the disease progresses, there is an increasing dependence on walking aids. Initially, patients depend on furniture, walls, and other people for support. Later on, they rely on canes, crutches, and wheeled walkers
- Truncal ataxia results in swaying when sitting and may necessitate back support

# A Closer Look at Common Symptoms of FA

(cont'd)



## Cardiac involvement

- While a majority of patients with FA will have evidence of cardiac complications (including cardiomyopathy and arrhythmias), patients are often asymptomatic. Palpitations are sometimes reported but overt symptoms of heart failure are uncommon<sup>2</sup>
- Heart disease in patients with FA can be severe and may contribute to disability and premature death. This is especially true in early-onset cases<sup>1</sup>
- Heart failure and arrhythmias (supraventricular in origin) are the most commonly reported causes of death<sup>1</sup>



## Diabetes mellitus/Hyperglycemia<sup>3,4</sup>

- When compared with age-matched populations, diabetes is more prevalent in patients with FA—with incidence estimates varying between 8% and 32%. Younger age at onset with longer disease duration places patients at an increased risk for diabetes



## Speech and swallowing

- Dysarthria is present in more than 90% of patients with FA. Over time, speech becomes slow and slurred, and patients become harder to understand in more advanced stages<sup>2</sup>
- As the disease progresses, dysphagia can become problematic, occasionally requiring gastroesophageal tube insertion. Patients may cough or choke on solids or liquids (including saliva), and chewing may be compromised. In some patients, this requires avoidance of tough foods, cutting food into small pieces, or increasing the bolus viscosity of liquids<sup>2,4</sup>



## Skeletal abnormalities

- When assessed clinically, scoliosis is present in approximately two-thirds of individuals with FA, a number that increases to 100% when assessed radiographically<sup>5</sup>
- Scoliosis is common early in FA—particularly when a patient has a poor recovery from scoliosis surgery or presents subtle neurological signs during or after surgery<sup>2</sup>
- The most rapid progression of scoliosis occurs between the ages of 10 and 16, corresponding to the age of puberty, and is associated with significant growth<sup>4,6</sup>
- Between 55% and 90% of patients with FA have foot deformities—including cases of both high arch and clubfoot. Clubfoot is a progressive condition found in advanced disease and can be very disabling with respect to mobility, transfers, and seating<sup>2</sup>



## Muscle weakness and wasting<sup>2</sup>

- Weakness occurs later in the course of the disease and is more prominent in the lower limbs compared with the upper limbs
  - Many patients preserve upper limb strength even when a wheelchair becomes necessary. Some patients may only ever develop mild distal upper limb weakness
- A significant number of patients experience wasting, and for patients who develop the disease in early life, muscle mass may never fully develop

# A Closer Look at Common Symptoms of FA

(cont'd)



## Ophthalmic features<sup>2</sup>

- Abnormal eye movement is a common early sign in the course of FA, with fixation instability being the most common feature. Nystagmus is less common but still frequent
- Decreased visual acuity is less common than eye movement abnormality, and the majority of patients are asymptomatic
  - On occasion, however, sudden bilateral loss of vision has been observed, mimicking Leber's hereditary optic atrophy



## Hearing<sup>2</sup>

- Most patients show disordered neural conduction in the central auditory pathways. This results in patients having trouble understanding speech in situations with everyday background noise



## Bladder<sup>2,5</sup>

- Symptoms of bladder hyperactivity are common in FA and are exacerbated by mobility problems



## Progression and mortality

- The mean duration of time from disease onset to use of a wheelchair is 15.5 years. On average, wheelchair use begins at 25 years of age<sup>2</sup>
- Symptoms such as dysarthria, lower limb pyramidal weakness, distal upper limb wasting, and loss of vibrational and joint position sense appear as the disease progresses<sup>2</sup>
- The largest retrospective study of mortality in FA included 61 individuals who had died<sup>2</sup>
  - Mean age at death was 36.5 years
  - Cardiac or probable cardiac dysfunction accounted for 62% of deaths. Of these, the majority resulted from heart failure or arrhythmia
- Survival into the sixth and seventh decades has been documented<sup>5</sup>

The table on the next page outlines the major clinical and genetic features that distinguish FA from other ataxias.



# Clinical and Genetic Features<sup>1,2,7-12</sup>

The information listed in the table below may be useful when trying to distinguish between the different types of ataxia.

Main clinical and genetic features of Friedreich's ataxia and other neurological disorders with similar clinical characteristics						
FEATURE	FRIEDREICH'S ATAXIA	ATAXIA TELANGIECTASIA	ATAXIA WITH OCULOMOTOR APRAXIA TYPE 1	ATAXIA WITH OCULOMOTOR APRAXIA TYPE 2	AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY	CHARCOT-MARIE-TOOTH DISEASE TYPE 1 (CMT1)*
<b>Usual age of onset</b>	<20 years (range, 2 years to >50 years)	<5 years (range, 2 years to 30 years)	<7 years (range, 2 years to young adult)	10-22 years	12-18 months (might occur later outside Québec)	5-25 years
<b>Cerebellar atrophy</b>	Present only in advanced cases	Present	Present	Present	Present	Absent (rare in Western world)
<b>Pyramidal signs</b>	Frequent	Present	Absent	Sometimes present	Present	Present
<b>Peripheral neuropathy</b>	Present (sensory axonal)	Present (axonal)	Present (motor and sensory axonal)	Present (motor and sensory axonal)	Present	Present
<b>Other signs and symptoms</b>	Kyphoscoliosis; pes cavus; optic atrophy; hearing difficulties; diabetes	Oculomotor apraxia; tremor; dystonia, telangiectasias of the conjunctiva; frequent sinopulmonary infections	Oculomotor apraxia; chorea; dystonia	Oculomotor apraxia; dystonia; chorea; tremor; cognitive impairment	Myelinated optic nerve fibers in the retina; scoliosis; pes cavus	Progressive distal muscle weakness; atrophy often associated with mild to moderate sensory loss; depressed tendon reflexes, bone deformities; pes cavus
<b>Cardiomyopathy</b>	Present	Absent	Absent	Absent	Absent (but mitral valve prolapse common)	Absent
<b>Gene and nature of mutations</b>	<i>FXN</i> ; <i>GAA</i> repeat expansion, rare point mutations (always in heterozygosity with <i>GAA</i> repeat expansion)	<i>ATM</i> ; nonsense mutations, frameshift, missense and leaky splice-site mutations, insertions and deletions	<i>APTX</i> ; missense, nonsense, frameshift and splice-site mutations	<i>SETX</i> ; loss-of-function missense, nonsense and truncating mutations; large-scale rearrangements	<i>SACS</i> ; missense mutations, deletions and insertions	<i>PMP22</i> ; duplication; point mutations

\*Inherited in an autosomal dominant pattern.

*FXN*, frataxin; *GAA*, glucosidase alpha, acid; *ATM*, ataxia-telangiectasia mutated; *APTX*, aprataxin; *SETX*, senataxin; *SACS*, saccsin; *PMP22*, peripheral myelin protein 22.

# Multidisciplinary Specialists

## Care Team Roles and Responsibilities

FA is a complex condition with variable clinical phenotypes that often require a broad multidisciplinary approach focusing on symptom management. Assembling the right care team will vary based on a patient's specific needs and circumstances.<sup>3</sup>

**Below is a list of specialists who may play a role in helping to provide specialized care to patients with FA.**



### Genetic counselor<sup>13</sup>

When considering an FA diagnosis, a geneticist can help patients undergo genetic testing for the disease and provide counseling to the patient and family, including discussions about risks for other family members or the patient, and what it could mean for family planning.

They also provide guidance to treating physicians in terms of ordering appropriate tests and helping interpret complicated test results.



### Primary care physician

The primary care physician provides consistent care for FA patients in all healthcare needs not directly related to FA. They can screen for FA complications, including cardiovascular issues, diabetes, and scoliosis.



### Cardiologist<sup>13</sup>

Patients with FA experience a high number of cardiac symptoms and can be diagnosed with cardiomyopathy. Clinical management guidelines recommend that a cardiologist perform an electrocardiogram and an echocardiogram at the time of diagnosis, then at least once a year and provide treatment as necessary. Due to the increased risk of arrhythmia, there is a potential need for care from a cardiac electrophysiologist.



### Endocrinologist<sup>13</sup>

The endocrinologist screens for glucose intolerance to establish a baseline in all patients with FA. They can also help counsel patients with impaired glucose tolerance or diabetes on the importance of lifestyle changes and prescribe treatment to control blood sugar levels, if necessary.



### Pulmonologist<sup>13,14</sup>

Pulmonologists can provide treatment options to help keep patients' lungs working optimally.



### Ophthalmologist/Audiologist

Ophthalmologists and audiologists can perform a comprehensive vision screening and auditory evaluation, and can provide tools and support to improve day-to-day hearing or visual issues.



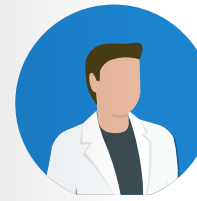
### Orthopedic surgeon<sup>13</sup>

Should orthopedic complications arise, orthopedic surgeons can help recommend the best course of action for musculoskeletal symptoms of patients with FA.



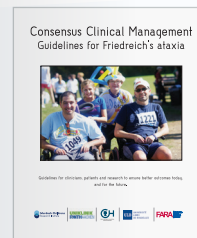
### Physical/Occupational therapist

Physical and occupational therapists can evaluate and optimize functional abilities and identify ways for patients to accomplish everyday tasks.



### Other specialists who can help your patients:

- Physicians
- Nutritionists
- Podiatrists
- Palliative care teams
- Speech therapists
- Social workers



### Learn more about managing patients with FA

Consensus clinical management guidelines for Friedreich's ataxia are available on the Friedreich's Ataxia Research Alliance (FARA) website. FARA is an organization focused on research and awareness for FA.

**To review the guidelines, visit [CureFA.org/clinical-care-guidelines](https://www.curefa.org/clinical-care-guidelines).**

## Get Connected to the FA Community

Despite there being a small number of FA patients across the country, various networks exist to bring together patients, clinicians, and researchers.



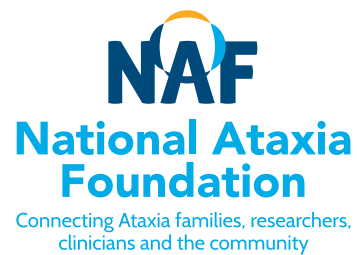
**Friedreich's Ataxia Research Alliance (FARA)** is dedicated to scientific research. The alliance raises funds for FA research, promotes public awareness, and brings together patients, clinicians, and other organizations with an FA-related focus. Research funded by FARA has led to a better understanding of gene mutation, frataxin production, iron sulfur cluster formation, and mitochondrial dysfunction.

To learn more, visit [CureFA.org](http://CureFA.org).



**Muscular Dystrophy Association (MDA)** is committed to improving the lives of people with muscular dystrophy and other neuromuscular diseases through innovations in science and care. FA is one of the 40 disorders addressed by the association. MDA's 230 hospital-affiliated clinics offer quality multidisciplinary care from doctors, nurses, and therapists experienced in dealing with neuromuscular diseases.

To learn more, visit [mda.org/care/mda-care-centers](http://mda.org/care/mda-care-centers).



**National Ataxia Foundation (NAF)** is dedicated to improving the lives of people living with ataxia through support, education, and research. At [ataxia.org](http://ataxia.org), there are free publications on FA management topics, such as the importance of exercise and the purpose of an ataxia diet.

To learn more, visit [ataxia.org](http://ataxia.org).

## Importance of Healthy Eating

A registered dietitian nutritionist can counsel patients on how food choices might promote health and better disease management.

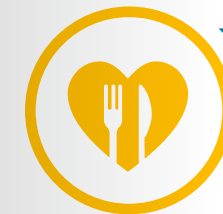
The recommended diet for most patients with ataxia is similar to what you might expect for general healthy eating. Please be mindful that between 8% and 32% of patients with FA also have diabetes and these patients will require extra dietary guidance not covered in this section.<sup>4</sup>



### Goals<sup>15</sup>

The goal of diet guidelines for ataxia is not to offer a cure for FA but rather to:

- Reduce the severity of some bothersome symptoms
- Reduce reliance on poorly tolerated or contraindicated pharmacotherapies
- Enhance the patient's perception of personal control and sense of responsibility for the management of his/her neurological condition



### Benefits<sup>15</sup>

Some of the benefits of an ataxia diet include:

- Sound nutrition to support healthy body weight and normal bodily functions. It is important to achieve an appropriate body weight for improved movement ability and lower stress on joints
- Increased energy and less fatigue
- High fiber may add regularity to bowel movements
- Improved mood and spirit



### High-fiber diet<sup>15</sup>

- Ataxia patients may benefit from a diet that restricts simple carbohydrates and is high in fiber. Ataxia patients may crave high-sugar foods; however, these foods may cause even more fatigue and depression than they relieve
- Certain patients may benefit from fiber supplements. Recommended dietary fiber intake is 30 to 40 grams a day; 15 grams is the norm for adults in the United States

## Role of Physical Activity<sup>15</sup>

While healthy eating is a great start, most patients with ataxia may also benefit from regular exercise. A physical therapist on your patient's care team ensures that they are instructed on exercises tailored to delay the advancement of balance problems.

# The Role of the mFARS

The primary goal of FA management is to help patients maintain their overall health and independence for as long as possible. The Modified Friedreich's Ataxia Rating Scale (mFARS) is a tool to help achieve this goal. The assessment provides a clinically meaningful way to track the progressive effects of FA on your patient's physical function and to anticipate the impact on their daily living. Because of their correlation with disease progression, changes to mFARS scores over time are typically accepted as an endpoint in clinical trials for potential new FA treatments.

DOMAIN	RELATED DAILY ACTIVITIES	ASSESSMENT GOALS	WHAT THIS MEANS FOR THE PATIENT
<b>Bulbar Function</b>	Swallowing or speaking	Measure strength and volume of coughing and clarity of speech <sup>16</sup>	<p>Reduced ability to cough brings greater risk of respiratory infection.<sup>17</sup> Loss of speech (dysarthria) impacts communication<sup>18</sup></p> <p><b>Care considerations:</b> Regular vaccinations, cough-assist devices, speech therapy</p>
<b>Upper Limb Coordination</b>	Brushing teeth, typing, pointing, reaching, turning a doorknob	Measure motor abilities related to tremors, fine motor coordination, and steadiness of hands and arms <sup>16</sup>	<p>Self-dressing and feeding may be affected, as well as handwriting and typing/texting.<sup>19</sup></p> <p><b>Care considerations:</b> Voice-based virtual assistant technology, occupational and/or physical therapy</p>
<b>Lower Limb Coordination</b>	Pressing foot pedals when driving	Measure coordination of legs and feet <sup>16</sup>	<p>Lower limb coordination closely relates to upright stability and is a strong indicator of disease progression.<sup>20</sup></p> <p><b>Care considerations:</b> Mobility aids (e.g., cane, walker, wheelchair); occupational and/or physical therapy</p>
<b>Upright Stability</b>	Walking, getting out of a chair, climbing the stairs, or showering	Measure loss of ambulation and impaired ability to coordinate voluntary movements <sup>16</sup>	<p>The course of disease for each patient is most evident in this domain. Over time, these scores help predict loss of function.<sup>20</sup></p> <p><b>Care considerations:</b> Mobility aids (e.g., cane, walker, wheelchair), occupational and/or physical therapy</p>

# Managing Your **FA** Patients

This brochure is designed as a resource for neurologists managing FA patients.

## What's inside:



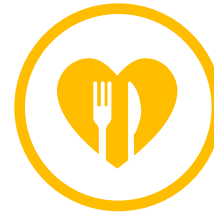
A closer look at common symptoms of FA



Detailing the multidisciplinary care team



Getting connected to the FA community



Discussing nutrition and exercise with patients

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