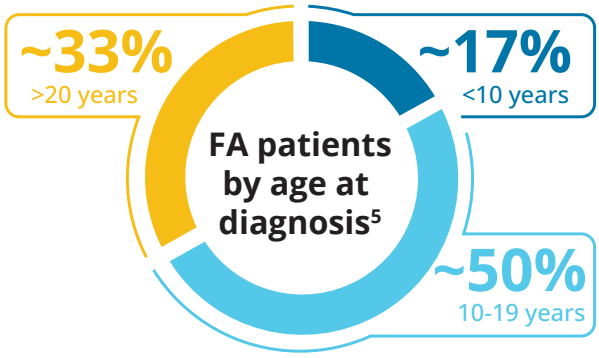
ThinkFA

Tricia, age 44
Diagnosed at age 29

Friedreich ataxia (FA) is a progressive, incapacitating neurodegenerative disease

FA is the most common form of inherited ataxia, affecting approximately 5000 or more people in the United States.[1,2] The condition results in **progressive loss** of functional abilities, leading to incapacitation and, eventually, death.[3,4]



Signs of FA typically appear between the ages of 10 and 15 years, but symptoms can emerge at **nearly any age**.[3]

Time is function for patients with FA, so think FA **FIRST**

Consider FA when you see any combination of these symptoms and shorten the time to diagnosis[6,7]:

Falls
(gait ataxia)

Imbalance
(poor proprioception)

Reflex loss
(areflexia)

Sensation loss
(neuropathy)

Tiredness
(fatigue)

Additional signs that may be present throughout the course of the disease:

- **Cardiomyopathy** may be present in younger patients with more severe FA[3]
- **Loss of bulbar function** may cause difficulty swallowing or slurred speech[3,8]
- **Scoliosis** may indicate FA when you also see other neurological signs[3]

FACT:

The most common form of inherited ataxia is commonly misdiagnosed

FA can present with **nonspecific or atypical symptoms**, making it common for the condition to be overlooked or misdiagnosed.[3,9] As many as 1 in 4 patients with FA have been misdiagnosed.[5]

Other inherited ataxias and conditions may overlap with symptoms of FA[3,5]:

- Spinocerebellar ataxia (SCA) - ICD-10 G11.2 & G11.8
- Ataxia-telangiectasia - ICD-10 G11.3
- Unspecified ataxia - ICD-10 G11.9 & G11.10
- Multiple sclerosis (MS) - ICD-10 G35
- Charcot-Marie-Tooth (CMT) disease - ICD-10 G60
- Cerebral palsy (CP) - ICD-10 G80.9

Any patient with a G11 diagnostic code can be considered for genetic testing to identify FA

Patients with an unconfirmed diagnosis of a hereditary ataxia may benefit from genetic testing

A genetic test that includes a GAA triplet-repeat expansion analysis is the only way to confirm an FA diagnosis[10]

Every potential FA patient should have their diagnosis confirmed with a genetic test, regardless of symptom presentation. But not all laboratories offer a genetic test that can detect the variant that causes FA, so ordering the right test from the right lab is critical.

Learn more about genetic testing for FA at ThinkFA.com

Sam, age 33
Diagnosed at age 15

Learn more about identifying and diagnosing patients with FA at ThinkFA.com

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