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Presentation and Progression of Friedreich Ataxia and Implications for Physical Therapist Examination

Joyce R Maring, Earllaine Croarkin

Friedreich ataxia, although rare, is the most prevalent inherited ataxia. Recent insight into the disease pathogenesis is creating new hope for effective therapies. The purposes of this update are: (1) to review the etiology, presentation, and progression of Friedreich ataxia and (2) to describe a comprehensive physical therapist examination emphasizing valid and reliable performance measurements associated with disease progression. Early identification of individuals with Friedreich ataxia and precise characterization of impairments and functional limitations gain importance as new drug therapies are considered.

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Presentation and Progression of Friedreich Ataxia

Friedreich ataxia (FRDA) is a progressive, autosomal recessive, degenerative disorder affecting multiple systems. The disease is named after Nicholas Friedreich, the physician who first described the condition in 1863. He described a degenerative atrophy of the posterior column of the spinal cord that caused progressive ataxia, sensory loss, and muscle weakness. The cardinal feature of FRDA is progressive gait and limb ataxia. Although rare, it is the most prevalent inherited ataxia, affecting about 1 in every 50,000 people, and it has an estimated carrier prevalence of 1 in 110 people.¹ More recent studies based on molecular data suggested that the prevalence may be as high as 1 in 29,000 people.² The genetic basis of FRDA was elucidated in 1997 and has led to a new understanding of its pathogenesis.^{3,4} With this enhanced understanding, a hope for the development of new therapies to slow the progression of and ultimately cure this fatal disease has evolved.⁵

Given that the cardinal feature of FRDA is gait and limb ataxia, a physical therapy evaluation to address identified concerns about an individual's poor balance or lack of coordination may well occur prior to genetic testing and a diagnosis of FRDA. However, because of the rel-

atively low prevalence of FRDA, people involved in the care of an individual with FRDA may be unfamiliar with the multisymptom presentation and progression of the disease. Few published resources are available to physical therapists to assist in recognizing and examining common signs and symptoms in people with FRDA. The need to recognize and accurately diagnose the disease in a timely way increases in importance as new therapies are being considered. The precise characterization of impairments and limitations with sensitive and well-validated tools has the potential to assist in the evaluation of the efficacy of new interventions under consideration.

The purposes of this article are to review FRDA etiology, pathology, and disease progression and to describe a comprehensive physical therapist examination for people with FRDA. Given the breadth of this topic and the lack of research on effective interventions for this population or similar populations, a detailed discussion of physical therapy treatments is beyond the scope of this article. However, we conclude with some general intervention recommendations based on typical examination findings and the available literature supporting intervention decisions for patients with FRDA as

well as other patient populations with degenerative or progressive neurological conditions.

Etiology and Pathogenesis

Friedreich ataxia results from an impairment in the FRDA gene, specifically, in the number of guanine-adenine-adenine (GAA) trinucleotide repeats. The majority of people who are healthy have 7 to 12 GAA repeats; in contrast, chromosomes in people with FRDA have 80 to 1,200 GAA repeats.² This altered expression leads to the partial loss of frataxin, a protein important in mitochondrial iron homeostasis.⁶ The amount of residual frataxin is inversely correlated with the number of GAA repeats.³ The loss of frataxin results in an accumulation of iron within mitochondria, which leads to excess production of free radicals. This excess production then results in cellular damage and death.⁷ The Figure summarizes this process. Tissues with a high energy demand are most susceptible to iron overload. In patients with FRDA, this condition is most evident in cardiac muscle, pancreatic islet cells, and the nervous system.^{3,8}

Neuromuscular System

Impairments in the neuromuscular system are essential clinical features of FRDA.^{7,9-11} The principal nervous

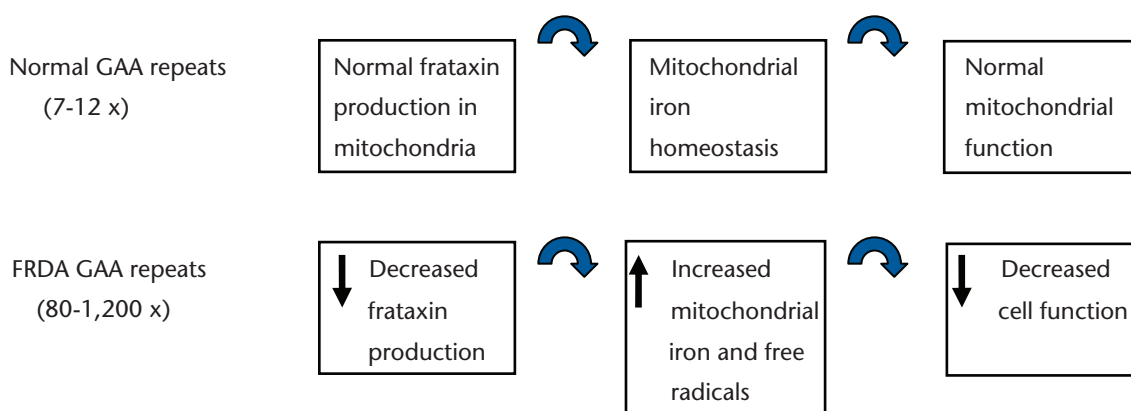


Figure.

Molecular etiology of Friedreich ataxia (FRDA). GAA=guanine-adenine-adenine trinucleotide.

system changes include a loss of cells in the dorsal root ganglia followed by degeneration in the posterior columns and spinocerebellar tracts. The deep nuclei and efferent pathways of the cerebellum and the corticospinal tracts also degenerate later in the disease process.¹²

The signs and symptoms of the disease have been well correlated with the degenerative changes that occur in the nervous system. The first sign to appear is usually gait ataxia.⁶ Lack of coordination and tremors develop in all extremities, with a progressive and symmetrical loss of strength (force-generating capacity). Tendon reflexes are diminished early in the disease process; spasticity and extensor plantar reflexes may emerge later in conjunction with corticospinal degeneration.⁶ In a longitudinal study of 33 patients with FRDA, participants exhibited a pattern of weakness affecting the pelvic girdle muscles first and then progressing in a variable fashion to other muscle groups of the lower extremities.¹³ The upper-extremity and trunk musculature was relatively spared until late in the disease process. The overall strength of the upper extremities was typically 80% of normal, and lower-limb strength averaged 70% of normal at the time when patients began to use a wheelchair for mobility. The authors concluded that weakness, although present, was not the most debilitating aspect of FRDA.¹³

People with FRDA frequently exhibit dysarthria, scanning speech, and dysphagia.¹⁴ Optic atrophy occurs in some patients and produces various degrees of visual loss. Oculomotor abnormalities include nystagmus, ocular flutter, and impairment of the vestibulo-ocular reflex. Hearing loss of the upper and lower frequencies has been reported and suspected to be related to spiral ganglion neurons, which carry auditory signals from

the inner ear hair cells to the brain.¹⁵ There is a gradual loss of light touch, position, and vibration senses in the extremities. The pattern of loss is typically distal and symmetrical.¹⁶

Cardiovascular System

Heart disease is a relatively common finding in FRDA. In a group of 115 patients diagnosed with FRDA, two thirds had abnormal electrocardiogram findings and 46 manifested symptoms of heart disease.¹⁷ People who were symptomatic complained of dyspnea on exertion and palpitations. In this cohort, abnormal electrocardiogram findings included a widespread T wave inversion. The primary cardiomyopathy-associated changes are fiber loss, diffuse fibrosis, focal myocardial necrosis, and cellular hypertrophy.¹⁷

In a more recent investigation of 29 patients with FRDA, myocardial velocity gradients were used to characterize the myocardium.¹⁸ Compared with age-matched control subjects, all of the participants demonstrated changes in the myocardium whether or not cardiac symptoms were present. The changes included increases in interventricular septal thickness, posterior left ventricular wall thickness, and left atrial diameter. These anatomical changes, along with the resultant effects of diminished mobility, can lead to inefficient cardiac function and impaired aerobic capacity.

Musculoskeletal System

The prevalence of scoliosis in people diagnosed with FRDA approaches 100%.^{19–23} People can exhibit a severe, progressive curve ($>60^\circ$), which should be treated surgically, or a less severe, nonprogressive curve ($<40^\circ$), which should be monitored.²² More progressive curves tend to occur before 10 years of age, whereas nonprogressive curves present during or after puberty. People who use a wheelchair are predis-

posed to developing a severe curvature.^{22,24} The type of scoliosis that occurs in patients with FRDA more closely resembles idiopathic scoliosis, which includes a more severe rotation component than scoliosis secondary to neuromuscular causes.^{22,25,26} Labelle et al²² identified no correlation between the degree of muscle weakness and the progression of scoliosis.

Malalignment of the feet has been considered to be a secondary sign of FRDA. Pes cavus can occur in up to 96% of patients diagnosed with FRDA.²⁷ However, pes planus or an equinovarus malalignment can also occur.^{28,29} If pes cavus or equinovarus is severe, mobility can be compromised secondary to the biomechanical disadvantage of a reduced or altered weight-bearing surface.

Endocrine System

Between 10% and 20% of patients with FRDA develop clinically apparent diabetes.³⁰ An additional 30% demonstrate impaired glucose tolerance.³¹ Virtually all patients with FRDA demonstrate a reduced early insulin response to intravenous arginine stimulation, a technique that measures beta cell secretion.³² Ristow et al³² demonstrated that non-insulin-dependent diabetes mellitus was strongly associated with the GAA trinucleotide repeats diagnostic of FRDA. These authors hypothesized that frataxin may be an important antioxidant and that the lack of frataxin may be involved in B-cell damage and other factors associated with non-insulin-dependent diabetes mellitus.

Medical Diagnosis and Prognosis

Although there are significant variations in the onset and rate of progression of the disease, the essential clinical criteria for the diagnosis of FRDA include an age of onset before 25 years, progressive ataxia, absent

knee and ankle jerk reflexes, and the presence of pyramidal signs.⁹ Disease symptoms typically appear between the ages of 5 and 15 years, with less frequent manifestations in early childhood. In rare cases, symptoms can appear as late as the third or fourth decade of life.³³ The mean (\pm SD) age of onset of symptoms of FRDA is 10.72 (\pm 7.4) years,³⁴ although one study³⁵ reported a mean onset as early as 6.1 years. The diagnosis of FRDA is confirmed by genetic testing when family history and clinical signs are consistent with the disease manifestations. The length of the GAA trinucleotide repeat is inversely correlated with the age of onset and provides a genetic marker of severity.³⁶

The symptoms associated with FRDA are progressive. Lack of coordination and ataxia, typically the first recognized symptoms, adversely and progressively affect the activities and participation of affected people. Patients with FRDA primarily use a wheelchair for mobility at an average age of 25 years, approximately 15.5 (\pm 7.4) years after disease onset.³⁴ Female patients progress more quickly to aided or wheelchair mobility than male patients with a comparable age of onset and disease severity.³⁷ Progressive worsening of motor symptoms is associated with degeneration of the cerebellar and corticospinal pathways.³⁸ Lack of coordination, ataxia, and balance impairments appear to be the major factors limiting mobility, as muscle strength is relatively preserved.^{13,22}

The mean age of death has been reported to be approximately 37.5 years, with a range as wide as 5 to 71 years.^{7,34} Death usually is secondary to progressive cardiomyopathy, pneumonia, or aspiration.²³ There are no reported sex differences in life expectancy, despite the fact that mobility function appears to

deteriorate more rapidly in female patients.³⁷

No known effective cure or treatment exists for FRDA. Intervention efforts currently focus on ameliorating symptoms and accompanying complications in order to assist patients in maintaining optimal function for as long as possible. Diabetes and heart problems can be treated with medications.

The discovery of the underlying genetic mechanism for FRDA has accelerated the understanding of its pathogenesis and has provided new hope for a cure. The efficacies of new drugs are currently under investigation.³⁹ These advances have generated the need to examine and validate outcome and performance measures that are meaningful and clinically sensitive to changes that can occur as a result of new therapies.

Physical Therapist Examination

Understanding the pathogenesis of FRDA and its multisystem involvement is critical in guiding a physical therapist's examination to lead to an accurate diagnosis and effective management decisions. The examination should consist of a complete history, a systems review, and the implementation of the best available tests and measures to describe a patient's impairments and functional limitations.⁴⁰ We review the components of the physical therapist's examination and make recommendations for best practice based on a compilation of typical impairments secondary to FRDA pathogenesis and, when available, the evidence described in the literature.

History

A systematic gathering of a comprehensive history is a critical feature of the physical therapy examination of patients with FRDA. Because the

clinical manifestations involve multiple systems, data accurately describing previous tests, surgeries, and interventions are important for understanding the disease progression and the complexity of involvement in an individual patient. For instance, given the high incidence of diabetes in this population, patients should be tested for glycosuria at approximately 6-month intervals.³⁰ Because many patients, families, and physicians are unfamiliar with this recommendation, a directed question during the family and patient interview may serve as a useful prompt.

Gaining insight into psychosocial factors also is essential in the interview process. The progressive nature of the symptoms of FRDA can influence an individual's perception of his or her quality of life to various degrees. D'Ambrosio et al⁴¹ surveyed 151 patients with hereditary ataxias and analyzed the relationships of the degree and type of disability to a patient's report on selected aspects of quality of life. The authors concluded that the degree of perceived independence and strong social and family support were important considerations in an individual's appraisal of life satisfaction. Comprehending the physical, social, and cultural contexts in which a patient functions is important for determining appropriate interventions that will facilitate the maintenance of meaningful activities and participation.

Systems Review

Because of the multisystem involvement of FRDA, the systems review should be comprehensive. Special attention should be directed to screening the physiological status of the cardiovascular system. Additionally, physical therapists should screen the musculoskeletal system for asymmetries frequently noted in people with FRDA, such as foot and

spine deformities. Because scoliosis is most likely to progress within the adolescent growth spurt,²⁸ screening should occur every 6 months during this period.

Tests and Measures

Selecting specific tests and measures to quantify and describe impairments and functional limitations as well as to inform interventions is a challenge with regard to examining patients with FRDA. In general, patients with FRDA may be classified under the preferred physical therapist practice pattern for impaired motor function and sensory integrity associated with progressive disorders of the central nervous system.⁴⁰ Many of the tests and measures listed in association with this practice pattern in the *Guide to Physical Therapist Practice*⁴⁰ are appropriate for quantifying or characterizing the signs and symptoms of the disease and disease progression. Below we highlight specific tests and measures that are important for examining patients with suspected FRDA or patients with a diagnosis of FRDA.

Traditional measurements of symmetry, range of motion, and muscle strength are good indicators of specific impairments of the musculoskeletal system. The prevalence of foot deformities and scoliosis in people with FRDA is very high,^{19-23,27} and these findings, in conjunction with impairments in motor control, should prompt the clinician to refer the patient for medical and genetic testing if such testing has not been performed. Range-of-motion limitations in other joints are not typical early in the disease process but may be common secondary to immobility and spasticity later in the disease progression.¹³ Muscle weakness is also not a significant finding early on; muscle strength may be fairly intact even when an individual is dependent on a wheelchair for mobility.¹³ When muscle weakness occurs, it is

more characteristic of the muscles of the pelvis and lower extremities than of the muscles of the trunk and upper extremities.

Cranial nerves should be tested for evidence of impairment of ocular movements, visual field and acuity deficits, hearing loss, dysarthria, and dysphagia.¹⁴ A complete test of sensory integrity should be performed because sensory neuropathy is often present, with position sense being lost throughout the extremities and the sensation of pain, temperature, and light touch being diminished distally in a symmetrical distribution.¹² Reflex integrity should be examined, with an emphasis on deep reflexes, tone scales, and superficial reflexes and reactions. A decrease in or an absence of lower-extremity deep tendon reflexes distinguishes FRDA from some of the other childhood ataxias.⁴² A positive Babinski reflex may indicate corticospinal tract involvement later in the disease progression.⁶

Physical therapists should consider using stationary ergometers for submaximal graded exercise testing to measure endurance and cardiovascular responses because balance concerns frequently limit participation in higher-level dynamic activities.⁴³

Although balance measurements have not been well documented for patients with FRDA, standard methods that correlate measurements with the risk of falling exist. For example, the Functional Reach Test has been shown to be a useful balance measure in a variety of age ranges, including the age range in which a diagnosis of FRDA is most likely.⁴⁴ Other tests, such as the Pediatric Clinical Test of Sensory Interaction for Balance,⁴⁵ the Pediatric Balance Scale,⁴⁶ the Timed "Up & Go" Test,⁴⁷ the Timed "Up and Down Stairs" Test,⁴⁸ and the measurement of static standing as a substest of the

Friedreich Ataxia Rating Scale (FARS),⁴⁹ may be appropriate for people diagnosed with FRDA. Consistent implementation of standardized balance tests for this population may provide documentation of disease progression as well as the effect of therapies. There is no evidence that such systematic documentation currently exists, and further research is needed to determine the most appropriate balance measures for this population.

Impairments in motor control such as ataxia have been historically based on subjective descriptions, which lack reliability and validity.⁴⁹ Because ataxia is considered to be the disease hallmark of FRDA, gross observations and anecdotal reporting severely limit the usefulness of an examination. Kinematic analyses that provide spatiotemporal descriptions of movement may provide precise measurements. Patients with ataxia have demonstrated lower peak velocities and prolonged movement durations even in single-joint movement measurements.⁵⁰ However, such analyses are not widely available to clinicians, and the relationship of these findings to performance limitations has not been demonstrated. A more accessible and precisely quantifiable measurement may be force control measurements of various muscle groups with a commonly available dynamometer in the isometric testing mode. Accuracy, as measured by the ratio of mean force to target force, and variability, as determined by the coefficient of variation, can be determined by asking people to maintain a target force for 30 seconds. This method of testing may prove to be a useful proxy measure of impairments in motor control secondary to ataxia in clinical trials of new interventions.⁵¹

Gait characteristics such as speed, symmetry, and level of independence are considered to be fairly

reliable indicators of disease progression.⁵⁶ *Ataxic gait* has been historically defined in physical therapy as a gait with a wide base.⁵² This definition has been challenged by a recent investigation that revealed no significant differences in the base of support between subjects who were healthy and patients who had ataxia.⁵³ The base of support during gait was more closely linked to biomechanical rather than neurological factors, and ataxic gait was better described by decreased velocity and gait asymmetries. Tests of velocity and independence in gait are easily accomplished and represent important functional measurements for people diagnosed with FRDA.⁵⁶

Composite Performance Measures

Composite rating scales for a variety of performance measures may improve the reliability and validity of measurements that are related to the assessment of clinical outcomes in patients with impairments in motor control such as ataxia.⁵⁶ Composite measures may include important performance indicators of function in several domains that may be affected by ataxia. Two such composite measurement tools are the International Cooperative Ataxia Rating Scale (ICARS)⁵⁴ and the FARS.⁵⁵ The ICARS subtests include measurements of posture and gait disturbances, kinetic functions, and speech and oculomotor functions. The ICARS discriminated mildly affected subjects from control subjects and demonstrated no ceiling or floor effects, indicating that it can be used to examine a range of ataxia severities as well as disease progression.⁵⁶ Test-retest reliability has been well established for the ICARS, with an intraclass correlation coefficient of .95.⁵⁶

The FARS was published within the last 2 years, and since then, investigators have been examining its test

psychometrics.^{55,57,58} Interrater reliability has been reported to have intraclass correlation coefficients of greater than .74.⁵⁵ The FARS subscales assess disease stage, activities of daily living, upper- and lower-limb coordination, bulbar and peripheral nerve function, balance, oral motor control, and gait speed. These subscales more closely reflect the typical presentation of FRDA than do the ICARS subtests, and scores on each of the subscales are significantly correlated with other measures of disease duration and disability.⁵⁸ Fahey et al⁵⁷ found the FARS to be more sensitive to changes in disease progression than the ICARS.

Other composite measures that have been used for patients with FRDA include the Ataxia Clinical Rating Scale,⁵⁹ the Functional Ataxia Scoring Scale,⁶⁰ the Inherited Ataxia Clinical Rating Scale,²⁷ the Inherited Ataxia Progression Scale,²⁷ and the Northwestern University Disability Scale.⁶¹ All of these instruments include domains of measurement that have face validity for patients with FRDA. However, there is minimal psychometric evidence to support their use in documenting disease progression in this population. The Ataxia Clinical Rating Scale and the Inherited Ataxia Clinical Rating Scale⁶² demonstrated a relationship between score and disease duration ($r=.64$ and $.69$, respectively).^{27,63} Rater agreement was described but not quantified when the Functional Ataxia Scoring Scale was used to assess the effects of amantadine hydrochloride treatment in patients with FRDA.⁶⁰ The Table summarizes the available evidence supporting the use of these composite measures.^{27,55-65}

Although the composite measures available at this time may provide the best available performance and outcome scores for patients with FRDA, they do not assess activity and participation domains as de-

finied by the *International Classification of Functioning, Disability and Health*.⁶⁶ Assessments of the end results of interventions would be more meaningful if they included outcomes specifically related to an individual's involvement in life situations. More work needs to be done to develop instruments that meet sensitivity needs but also are geared to measuring meaningful changes.⁶⁷

Physical Therapy Intervention

Rehabilitation therapies usually focus on strategies and compensatory techniques for maintaining or improving a patient's ability to continue to participate in all environmental contexts for as long as possible. Physical therapists provide education to patients and family members about the effects of disease progression on function and lifestyle, potential therapeutic interventions, and realistic expectations regarding those interventions. At present, there is little evidence supporting specific physical therapy interventions that would address impairments or functional concerns in patients with FRDA.

Patients with FRDA may improve aerobic fitness by participating in stationary cycling for 20 to 25 minutes at 70% to 85% of their maximum heart rate, as measured with a graded exercise test.⁴³ To compensate for impaired upright stability, therapists should recommend that patients use a stationary bicycle or a stationary ergometer for aerobic conditioning. The benefits of physical exercise programs have been demonstrated for other patient populations with degenerative disabilities that include ataxia,^{68,69} and it is reasonable to assume that the health and function of people with FRDA also would be facilitated by regular participation in an adapted exercise program. Physical therapists can assist an individual with FRDA in identifying community

Table.
Composite Tests, Domains, and Test Psychometrics^a

Assessment	Domains Included	Test Psychometrics	Results of Analysis
International Cooperative Ataxia Rating Scale (ICARS) ⁵⁴	Gait, balance, coordination, speech, writing, oculomotor	Interrater reliability	Kendall correlation = .994, $P < .00001$ ⁶⁴ ICC = .95 ⁶⁵
		Internal consistency	Cronbach alpha = .94 ⁵⁶ Cronbach alpha = .95 ⁶⁵
		Test-retest reliability	ICC = .95 ⁵⁶ ICC = .97 ⁶⁵
		Construct validity of total ICARS assessment	Correlated with disease duration, $r = .61$ ⁵⁶ Correlated with age, $r = .41$ ⁵⁶ Correlated with disease stage, $P < .001$ ⁶⁵ Correlated with Barthel Index, $r = -.70$, $P < .0001$ ⁶⁵ Correlated with disease duration, $r = .43$, $P < .0001$ ⁶⁵
		Construct validity of ICARS subscales	Intercorrelations of ICARS subscales, $r = .21-.87$ ⁵⁶
Ataxia Clinical Rating Scale ⁵⁹	Cranial nerves, coordination, tone, reflexes, strength	Construct validity	Correlated with disease duration, $r = .64$, $P < .005$ ⁶³
Friedreich Ataxia Rating Scale (FARS) ⁵⁵	Disease stage, activities of daily living, limb coordination, bulbar and peripheral nerve function, oral motor control, gait speed	Interrater reliability	ICC > .74 ⁵⁵
		Construct validity of total FARS assessment	Correlated with modified Barthel Index, $r = .9$, $P < .001$ ⁵⁷ Correlated with Functional Independence Measure, $r = .94$, $P < .001$ ⁵⁷ Correlated with ICARS, $r = .96$, $P < .001$ ⁵⁷ Correlated with disease duration, $r = .79$, $P < .001$ ⁵⁷ Correlated with disease duration, $r = .52$, $P = .0014$ ⁵⁸ Correlated with activities of daily living and disease stage, $r = -.21-.90$ ⁵⁵
		Construct validity of FARS subscales	Correlations between FARS subscales and modified Barthel Index, Functional Independence Measure, and ICARS; $r = .87-.97$, $P < .001$ ⁵⁷ Correlations between FARS subscales and disease duration, activities of daily living, and disability; $r = .42-.92$ ⁵⁸
		Sensitivity to change	Effect size larger than those of modified Barthel Index, Functional Independence Measure, and ICARS ⁵⁷
Functional Ataxia Scoring Scale ⁶⁰	Oculomotor, speech, writing, coordination, trunk stability, scoliosis, cardiomyopathy	Rater "agreement"	$P < .005$ (no statistical methods reported) ⁶⁰
Inherited Ataxia Progression Scale ²⁷	Disability stages defined	None	
Inherited Ataxia Clinical Rating Scale ⁶²	Speech, tremor, coordination, balance, tone, vibration sense	Construct validity	Correlated with age, $r = .6987$, $P < .001$ ²⁷
Northwestern University Disability Scale ⁶¹	Walking, activities of daily living, speech	None	

^a ICC=intraclass correlation coefficient.

fitness and recreational activities that are most appropriate given the individual's specific impairments.

Maintaining biomechanical alignment is an important therapeutic consideration. Orthopedic problems such as foot deformities and scoliosis are often treated with orthoses or surgery and may result in a temporary improvement in function.²⁹ Early intervention for biomechanical changes in the foot significantly improves alignment and thus weight-bearing ability and mobility outcomes.²⁹ The plan of care should include daily attention to range of motion, including muscle length and soft-tissue extensibility. This plan of care likely would involve a home exercise program and family instruction for follow-through.

Maintaining independence in mobility is another area in which physical therapy consultation and intervention are warranted. Physical therapists are frequently involved in decisions regarding assistive or adaptive devices to be used during functional activities. The use of a reverse-brake-system walker has been reported to reduce fall frequency when a walker is indicated for maintaining independence in gait.⁵¹ However, some people with FRDA rely on lateral stepping strategies to prevent falling; for these people, walkers may reduce ambulatory safety.⁷⁰ When a wheelchair is indicated as a primary means of mobility, therapists may encourage patients and families to initially consider a power scooter for community transport and a manual wheelchair for inaccessible areas encountered in the usual environment. Similar to the assessment of wheelchair needs of children with muscular dystrophy, a powered wheelchair is indicated when a patient relies on the arm rests for trunk control or demonstrates severely asymmetrical sitting postures.⁷¹

Summary

Friedreich ataxia is a severe neurodegenerative condition, and new testing methods indicate that the disease may be twice as prevalent as previously thought.² Although at present FRDA is incurable, recent identification of the genetic mechanism and pathogenesis has provided new optimism for a disease-altering therapy. Current studies are investigating the effects of new drugs. People with FRDA may be referred for physical therapy prior to genetic testing and diagnosis. Physical therapists need to become familiar with the multisystem disease presentation in order to make appropriate clinical decisions, which may include a referral for genetic testing and medical diagnosis. Additionally, physical therapists need to investigate and validate outcome and performance measures that are clinically sensitive to changes that may occur with new drug interventions as well as physical therapy interventions.

Comprehensive interviews and systems reviews are important for evaluating the clinical manifestations and impact of this disease, which affects multiple systems. Discrete tests and measures should be consistently and reliably implemented. Composite measures may represent the best possible primary outcome measures at this time for quantifying the impact of motor control impairments on a variety of performance measures. Future test development should also consider measurements of functional changes related to an individual's participation in life situations. Given the current progressive nature of this disease, physical therapists should intervene with the goal of maintaining an individual's independent participation in all environmental contexts for as long as possible. Future research is necessary to determine which interventions can reduce impairments and improve

functional outcomes in patients with FRDA.

Both authors provided concept/idea/project design and writing.

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