Clinical Management Guidelines for Friedreich Ataxia

Chapter 9. Orthopedic issues in Friedreich ataxia

Contents

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9. Orthopedic issues in Friedreich ataxia

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This chapter describes orthopedic issues associated with Friedreich ataxia, specifically scoliosis and foot deformities, and strategies for investigating and managing these issues. In making recommendations for management of orthopedic issues, the authors were tasked with answering the following questions:

For individuals with Friedreich ataxia, what management strategies could be implemented for scoliosis? (see 9.2)

For individuals with Friedreich ataxia, what management strategies could be implemented for foot deformity? (see 9.3)

9.1 Overview of orthopedic issues in Friedreich ataxia

Orthopedic issues are apparent in Friedreich ataxia (FRDA) particularly in the majority as scoliosis and to a lesser extent, pes cavus (1, 2). In early (age 0 to 7 years) and typical onset (8 to 14 years) FRDA, 69% and 80% of patients respectively present with scoliosis (3). The majority of studies report the most rapid progression of scoliosis occurring between the ages of 10 and 16 years corresponding to the age of puberty and associated with significant growth (3). In a large European natural history study (n=649) deformities of the feet, predominantly pes cavus, was reported in 374/649 (58.8%) individuals. Of this cohort 47/372 (7.2%) required surgery. Both the presence of scoliosis and pes cavus have the potential to impact quality of life in terms of functional capacity and pain. This chapter discusses the orthopedic issues related to FRDA, in particular management of scoliosis and foot deformities.

9.1.1 Scoliosis in Friedreich ataxia

Scoliosis occurs in most individuals with FRDA (4-7), with a high prevalence of double thoracic and/or lumbar curves. Scoliosis may be severe and progressive, that is, occurring before the age of 10 years and exhibiting a curve greater than 60^{o}; or less severe and non-progressive with a curve less than 40^{o} (7). A large natural history study comprising 1116 individuals with FRDA reported intermediate to severe scoliosis in 90% of individuals with early (age 0 to 7 years) and typical (8 to 14 years) FRDA onset. There was a lower prevalence of scoliosis in those with a later age of onset (>14 years) (3). In a further large natural history study (n=650) scoliosis was reported in 73.5% of individuals with FRDA (2). A smaller prospective monocentric study reported 47/66 (71%) individuals with FRDA had a scoliotic deformity (8). The growth phase and puberty present times of major progression of scoliosis, resulting in the need for surgical intervention in more than 50% of individuals, on average aged between 12 and 16 years (3, 4, 9). More severe scoliosis is generally found in individuals who have an onset of FRDA prior to 10 years of age (3, 5, 7, 8, 10). An earlier age of onset of FRDA generally coincides with the rapid progression of scoliosis between the ages of 10 and 16 years; hence, this time is particularly critical in terms of monitoring and intervention (3). It should be noted that scoliosis related to FRDA does not always progress and therefore does not always require intervention (7, 11, 12).

Allard and colleagues (9) reported that typically an individual with FRDA presents between the ages of 10 and 15 years with a right thoracic spinal deformity which is relatively stable, usually with a Cobb angle of 24^{o}, however, thereby increasing in the following five years in addition to developing a

compensatory left lumbar curve. Subsequent studies reported scoliosis in FRDA may be a single thoracic curve; however, the more usual pattern is of double major curves (4, 6, 13).

The etiology of scoliosis in FRDA is still unclear. It does not follow the usual pattern associated with scoliosis of a neuromuscular origin, such as muscular dystrophy or spinal muscular atrophy (7, 8, 14). Muscle weakness, age of onset or clinical severity has not been shown to correlate with severity or progression of scoliosis in FRDA (7, 8). Spasticity is a significant component of the motor pattern disturbance associated with FRDA and it is possible that spasticity and/or disturbed postural reflexes may have a role in the development of scoliosis associated with FRDA. This hypothesis, while yet to be confirmed, may be significant in considering non-invasive methods of managing scoliosis in FRDA.

9.1.2 Foot deformity in Friedreich ataxia

Individuals with FRDA may present with pes cavus and also equinovarus deformities, both of which may place individuals with FRDA at a particular mechanical disadvantage and make locomotion, transfers and standing difficult (15-17).

9.2. Management strategies for scoliosis in Friedreich ataxia

9.2.1 Investigation of scoliosis

The usual method of investigation of scoliosis is antero-posterior and lateral full spine radiographs either standing or supine. However, Allard and colleagues (9) suggest this may lead to inconsistent measurement and recommends the use of a rotational booth. The Cobb method (18) is used to determine the degree of curve. Pelvic obliquity is defined as the angle between a line joining both iliac crests and a line intersecting the middle of the pedicles of L4 and L5 on the posteroanterior view (7, 19).

9.2.2 Interventions for scoliosis

A literature search from 1957 to 2021 revealed 14 articles related to management of scoliosis in Friedreich ataxia. The majority of these studies are retrospective, with sample sizes ranging from 1 to 78 (see Table 9.1 for details of each study).

Table 9.1: Summary of studies related to management of scoliosis in Friedreich ataxia

Author(s), Year	Type of study	Number of participants	Intervention	Outcome
Allard et al, 1982 (9)	3-year retrospective	32	Examination of pathomechanics of spinal deformity in FRDA.	Recommendation of examination and intervention.
Cady et al, 1984 (4)	Retrospective, case series (pre-post)	42	Non-operative and surgery.	Bracing not effective. Surgery is indicated for curves >40^{o}.
Daher et al, 1985 (13)	Retrospective, case series (pre-post)	19	Bracing and surgery.	Bracing not effective. Surgery is indicated for curves >40^{o}.

Author(s), Year	Type of study	Number of participants	Intervention	Outcome
Labelle et al, 1986 (7)	Retrospective	78	Non-operative and surgery.	Observation of curves <40°; observation and surgical treatment for curves >40° and <60°; surgical treatment for curves >60°.
Garfin et al, 1988 (11)	Retrospective	21	Bracing and surgery.	Bracing not effective. Surgery is indicated for curves with documented progression.
Shapiro et al, 1993 (5)	Literature Review		Bracing and surgery	Bracing for ambulant individuals with curve >25° and <35°. Consider surgery for non-ambulant individuals with curve>40°.
Aronsson et al, 1994 (14)	Retrospective	12	Comparison of shape of curve between children with cerebral palsy, FRDA and adolescent idiopathic scoliosis.	Curve of FRDA resembles that of adolescent idiopathic scoliosis and is not related to muscle weakness.
Stricker et al, 1996 (20)	Retrospective, case series (pre-post)	5	Posterior fusion with a modified Luque technique.	Significant complications with FRDA. Rigid fixation and immediate post-operative mobilization particularly important. Recommend two-step ventral and dorsal procedure when curve is rigid and >50°.
Milbrandt et al, 2008 (6)	Retrospective, case series (pre-post)	49	Spinal fusion (n= 16) and bracing (n=10).	Poor results with bracing. Fusion effective.
Piazzolla et al, 2011 (19)	Retrospective, case series (pre-post)	1	Cotrel-Dubousset instrumentation.	Good correction of deformity.

Author(s), Year	Type of study	Number of participants	Intervention	Outcome
La Rosa et al, 2011 (21)	Retrospective, case series (pre-post)	2	Cotrel-Dubousset instrumentation.	Good correction of deformity.
Tsirikos et al, 2012 (12)	Retrospective	31	No intervention, bracing and surgery.	Bracing may be useful in delaying surgical correction in young child. Posterior spinal fusion achieves and maintains good correction with minimal complications.
Simon et al, 2019 (8)	Prospective monocentric study	66	Bracing and posterior fusion	Brace success rate was 61%, but 19% of the cohort underwent posterior fusion, extended to the sacrum in non-ambulatory patients
Cotrel & Dubousset, 2014 (22)	Retrospective	3	Reports a new method of instrumentation aimed at obtaining surgical fixation of the scoliotic curve without any postoperative external immobilization	Report success of this type of fixator including being more rigid than the Harrington or Luque Rods, no neurological deficit related to surgery, good correction without need for bracing.

The goal of intervention in individuals with FRDA is to prevent progression of scoliosis, improve posture and the capacity for upright stability in those who are ambulatory, improve balance, and reduce the reliance on external orthoses for postural control and pressure/pain relief in those who are non-ambulatory (8). In addition, prevention of secondary respiratory complications is a significant factor to consider in the management of scoliosis in FRDA (19). In both ambulatory and non-ambulatory groups, pain is a significant indicator for intervention and thus successful intervention may have a significant impact on quality of life for people with FRDA.

Most authors recommend observation of children with a curve between 20^{o} and 40^{o}; however, they suggest that if the curve progresses beyond 40^{o}, intervention is indicated (4, 7). Intervention can be non-invasive, such as the use of bracing in the ambulatory individual or the use of customized seating to limit progression of rotation and lateral deviation of the spine in non-ambulatory individuals. Although the efficacy of bracing in controlling deformity has been questioned (6), bracing may be particularly valuable in the very young and compliant person with FRDA to slow

down progression and delay surgical correction (12). Unfortunately, bracing may interfere with ambulation and breathing and thus compliance may be an issue (4, 6, 9, 13).

Consideration of surgical intervention for scoliosis should take into account the negative effects of Implant failure, risk of infection and risk of loss of mobility in the recovery phase post-operatively (8). The fusion level (e.g., extending fusion to the pelvis), in particular the mobility status and projected loss of ambulation of each individual, should also be considered. Finally, the risk of intraoperative complications such as fluid loss/overload and associated cardiac issues need to be considered (8). Simon and colleagues (8) reviewed the rate of scoliosis progression in a small cohort of 28 individuals with FRDA who were managed conservatively and noted conservative treatment such as bracing did not influence the per-year rate of progression. Whilst there is little literature reporting post-operative mobility, Simon and colleagues (8) did report those who were still ambulatory when undergoing surgical intervention (n=5) were still able to walk two to five years postoperatively. As indicated by Rummey and colleagues (3), loss of ambulation often coincides with the requirement for scoliosis surgery, which confounds interpretation of surgical outcomes in regards to ambulation status.

Posterior fusion may be considered for progressive curves greater than 40^{o} in the child who is still growing (4, 7, 9). Milbrandt and colleagues (6) recommend consideration of surgery when the curves approach 50^{o}. Stricker and colleagues (20) expanded the criteria to include those individuals who experience significant loss of balance while sitting, poor control of the head and difficulty complying with orthotic devices. Piazzolla and colleagues (19) proposed that indications for surgery should include deformity causing functional problems (poor sitting balance, difficulties with hygiene), or impingement of the rib cage on the pelvis or a deformity that could progress to create any of all of these problems. It has been recommended that the fusion area extend from T2 to L3 or L4 and the instrumentation contoured to accommodate the usual thoracic kyphosis and lumbar lordosis (5, 6). However, some individuals may benefit from a shorter fusion and those who are non-ambulatory may require a fusion to the pelvis. The decision to fuse and the nature of the fusion should be based on each individual case taking into consideration the severity of the deformity, mobility and associated risks (12).

Spinal deformities are corrected using a posterior instrumented spinal fusion with pedicle screw fixation. In some cases, pelvic fixation may be required. Standard, modern intraoperative neuromonitoring may not be possible in all children with FRDA who need spine deformity correction, so the surgical team must be prepared to perform a wake-up test.

Cardiopulmonary assessment is essential prior to consideration of surgery to ensure cardiac function is not compromised during the procedure and in the post-operative phase (23).

See Chapter 4 for assessment and management of cardiac issues.

It is important to delay surgery for as long as possible by use of conservative management. When the scoliosis has progressed in terms of curve magnitude, surgery should be considered after careful multidisciplinary evaluation (including cardiac function). In consideration of surgical management of scoliosis for individuals with FRDA, it is essential to consider issues related to complications and post-operative management. In addition, it is important to consider if, depending on the ambulation status of the patient, the fusion should be extended to the pelvis. Some studies reported post-operatively immobilization in orthoses for periods from 3 to 15 months (4, 13). Moreover, significant intraoperative blood loss may compound pre-existing cardiopulmonary issues (23); hence it is critical surgery is conducted in a facility with an intensive care unit (ICU) for initial post-operative care. Complications related to loss of mobility, de-conditioning, infection, failure of the implants,

cardiopulmonary issues or death are reported in most studies. Post-operative requirements and availability in terms of acute community-based care and impact on life roles of the individual with FRDA (e.g., time off school, increase in level of dependency, possible deconditioning associated with post-operative recovery) need to be considered on a case-by-case basis. While there is some evidence that physical therapy may slow the progression and/or improve Cobb angles in idiopathic scoliosis (24), there is no evidence that such intervention would be effective in individuals with FRDA.

9.2.3 Conclusion

The available literature largely supports the role of surgery in correcting scoliosis in individuals with FRDA who demonstrate a curve >40^{o} and in whom presence of a curve has a significant impact on health, independence, and quality of life. However, expert authors consider it is important to delay surgery for as long as possible by the use of conservative management. Bracing, while not proven to affect the prognosis of the deformity or the need for surgical correction, may be valuable in delaying surgical correction in the very young child. When the scoliosis has progressed in terms of Cobb angle and associated discomfort, surgery should be considered only after extensive pre-operative multidisciplinary evaluation, with particular attention to cardiorespiratory function. If surgery is considered, the need to extend fusion to the pelvis (depending on ambulation status of patient) should be assessed.

Best practice statements

Spinal examination must be part of the multidisciplinary approach for individuals with Friedreich ataxia and should be performed regularly.
Individuals with Friedreich ataxia with a spinal curve between 20^{o} and 40^{o} and/or between the ages of 10 and 16 years should be observed for curve progression.
Bracing may not reduce or stop the progression of curves; however, it may be valuable in delaying surgical correction in the young child.
People with Friedreich ataxia with a scoliosis >40^{o} may be considered appropriate for surgical correction.
Consideration should be given to delaying surgical intervention in ambulant individuals with Friedreich ataxia.
All people with Friedreich ataxia considered for scoliosis surgery require extensive pre-operative evaluation and planning regarding cardiac and pulmonary function.

Recommendations

Grading for strength of recommendation and level of evidence

For the rating of the **strength** of the recommendation, in addition to evidence from studies in FRDA, evidence from like conditions, clinical experience and expert consensus are taken into account when published evidence is not available.

The **level of evidence** is based on published evidence from studies in FRDA. If there is no published evidence in FRDA, evidence from other like conditions or clinical expertise may have been used to make the recommendation – this is graded as ‘very low’ or in some cases ‘low’ level evidence. See the table below for an explanation of the symbols used to grade recommendations.

Strength of recommendation	Symbol	Level of evidence	Symbol
Strong for intervention	↑↑	High	⊕⊕⊕⊕
Conditional for intervention	↑	Moderate	⊕⊕⊕◯
Neither intervention nor comparison	—	Low	⊕⊕◯◯
Conditional against intervention	↓	Very low	⊕◯◯◯
Strong against intervention	↓↓		

Surgery for scoliosis

Should surgery versus conservative therapy be used for scoliosis with Friedreich ataxia?	**Strength**	**Level of evidence**
We suggest surgery should *not* be used over conservative therapy for individuals with scoliosis and Friedreich ataxia. Expert opinion suggests that conservative therapy, including the use of bracing during the time of rapid growth in children/adolescents (age 10 to 16 years), may assist in avoiding or delaying surgery.	↓	⊕◯◯◯
Justification: The expert authors consider it is important to delay surgery for scoliosis in individuals with Friedreich ataxia for as long as possible by use of conservative management. When the scoliosis has progressed in terms of Cobb angle and associated discomfort, surgery should be considered only after careful multidisciplinary evaluation (including cardiac function). If surgery is considered, the necessity or not to extend fusion to the pelvis should be assessed, depending on the ambulation status of patient.		
Subgroup considerations: This recommendation is for individuals with Friedreich ataxia with scoliosis. Ambulatory individuals with scoliosis are at risk of losing ambulation through the process of surgical intervention; therefore, those who are still ambulant may require particular consideration.		

Age and scoliosis surgery

Should age be considered versus none be used for scoliosis surgery with Friedreich ataxia?	**Strength**	**Level of evidence**
We recommend that age should be considered when contemplating scoliosis surgery for individuals with Friedreich ataxia.	↑↑	⊕◯◯◯
Justification: Expert opinion suggests that the use of bracing during the time of rapid growth in children with scoliosis (age 10 to 16 years) may assist in avoiding surgery. For young children (before age 10 years), we recommend aiming to prevent progression of scoliosis with bracing rather than surgery, in an effort to avoid early spine fusion.		
Subgroup considerations: This recommendation is for individuals with Friedreich ataxia with scoliosis. Younger individuals require specific attention when considering scoliosis surgery or other treatments.		

Cardiac status and scoliosis surgery

Should cardiac status versus none be used for stratifying scoliosis surgery with Friedreich ataxia?	**Strength**	**Level of evidence**
We recommend stratifying individuals with Friedreich ataxia according to cardiac status when considering scoliosis surgery.	↑↑	⊕◯◯◯
Justification: Individuals with Friedreich ataxia and concurrent cardiac disease may be at greater risk of intra and post-operative complications related to fluid loss/overload and congestive cardiac failure. Individuals with Friedreich ataxia undergoing scoliosis surgery should undergo EKG, echocardiography and a cardiology consultation within a reasonable time frame prior to the surgery (e.g., 2 to 4 months prior) to evaluate left ventricular (LV) structure and left ventricular ejection fraction (LVEF). The surgical plan should incorporate a cardiac management plan in conjunction with the cardiologist.		
Subgroup considerations: This recommendation is for Individuals with Friedreich ataxia who require scoliosis surgery.		

Lay summary

Lay summary of clinical recommendations for scoliosis in Friedreich ataxia

Why these recommendations?

Scoliosis (curvature of the spine) occurs in most individuals with Friedreich ataxia. Scoliosis can be severe, where it happens at a young age (earlier than 10 years of age) and gets progressively worse over time, or it can be less severe and not get much worse over time.

These recommendations suggest that individuals with Friedreich ataxia should have a regular examination of the spine, particularly young people aged between 10 and 16 years. If a spinal curve is apparent, it should be observed over time to see if it gets worse.

The options for correcting a scoliosis are either to wear an external brace or undergo surgery. For young children (before age 10 years), we recommend aiming to prevent progression of scoliosis with bracing in the first instance, in an effort to avoid early spine fusion with surgery. While bracing may not reduce or stop the progression of the curve, it may be valuable in delaying surgery, particularly for young children or someone who is still able to walk.

If surgery for scoliosis is being considered, individuals with Friedreich ataxia and cardiac disease may be at greater risk of complications related to fluid loss/overload and congestive cardiac failure during or after surgery. Individuals with Friedreich ataxia undergoing scoliosis surgery should therefore have an EKG, echocardiography and a cardiology consultation within a reasonable time before the surgery.

What does this mean for you as a person living with Friedreich ataxia or caring for someone living with Friedreich ataxia?

It might be important for you to speak with your healthcare professional about Friedreich ataxia and management of spinal curves and what it means for you.

Who are these recommendations specifically for?

These recommendations are specifically for individuals with Friedreich ataxia and scoliosis.

9.3 Management strategies for foot deformity in Friedreich ataxia

9.3.1 Surgery

There is little evidence to support the efficacy of orthopedic surgery for individuals with foot issues with Friedreich ataxia; however, expert opinion suggests surgery may be considered if it would assist in foot alignment to facilitate mobility and/or safe and effective transfers. In weighing up the advantages and disadvantages of foot surgery, the risk of infection, pain associated with the procedure, risk of loss of mobility in the post-operative recovery phase, sensory issues in managing post-operative casts/immobilization (if indicated), and the risk of intraoperative complications such as fluid loss/overload and associated cardiac issues should be considered.

9.3.2 Ankle foot orthotics

Previous guidelines indicate that ankle foot orthotics (AFOs) may be appropriate for people with FRDA to provide mediolateral stability at the ankle in stance phase, facilitate toe clearance in swing phase, promote heel strike at initial contact, prevent foot deformity, support normal joint alignment and biomechanics, improve range of motion and to facilitate function (25, 26).

See Chapters 3.2 and 3.3 for more details.

There are no studies specifically looking at orthotic prescription for people with FRDA; however, there are multiple studies describing the benefits of AFO prescription in improving postural security and movement, increasing weight-bearing percentage, improving knee control, reducing toe clawing, improving gait parameters, and improving standing and balance in individuals with other neurological conditions, such as stroke and spastic cerebral palsy (25-30).

Recommendations

Surgery for foot deformity

Should orthopedic surgery versus nothing be used for individuals with foot issues with Friedreich ataxia?	**Strength**	**Level of evidence**
We conditionally recommend orthopedic surgery for individuals with foot issues with Friedreich ataxia if it is considered such surgery would assist in foot alignment to facilitate mobility and/or safe and effective transfers.	↑	⊕◯◯◯
Justification: There is little evidence to support the efficacy of orthopedic surgery for individuals with foot issues with Friedreich ataxia; however, expert opinion suggests surgery may be considered if it would assist in foot alignment to facilitate mobility and/or safe and effective transfers. In weighing up the advantages and disadvantages of foot surgery, the risk of infection, pain associated with the procedure, risk of loss of mobility in the post-operative recovery phase, sensory issues in managing post-operative casts/immobilization (if indicated), and the risk of intraoperative complications such as fluid loss/overload and associated cardiac issues should be considered.		
Subgroup considerations: This recommendation is for individuals with Friedreich ataxia with foot deformity.		

Orthotics for foot deformity

Should orthotics (including ankle-foot-orthotics, ankle braces and in-shoe orthotics) versus nothing be used for individuals with foot issues with Friedreich ataxia?	**Strength**	**Level of evidence**
We conditionally recommend the use of orthotics (including ankle-foot orthotics, ankle braces and in-shoe orthotics) for individuals with foot issues with Friedreich ataxia.	↑	⊕◯◯◯
Justification: Ankle-foot orthotics (AFOs) may be appropriate for people with Friedreich ataxia to provide mediolateral stability at the ankle in stance phase, facilitate toe clearance in swing phase, promote heel strike at initial contact, prevent foot deformity, support normal joint alignment and biomechanics, improve range of motion, and to facilitate function (25, 26). There are no studies specifically looking at orthotic prescription for people with Friedreich ataxia; however, there are multiple studies describing the benefits of AFO prescription in improving postural security and movement, increasing weight-bearing percentage, improving knee control, reducing toe clawing, improving gait parameters, and improving standing and balance in individuals with other neurological conditions, such as stroke and spastic cerebral palsy (25-30).		
Subgroup considerations: This recommendation is the Individuals with Friedreich ataxia with foot issues.		

Physiotherapy for foot deformity

Should physiotherapy (including stretching, soft-tissue work) versus nothing be used for individuals with foot issues with Friedreich ataxia?	**Strength**	**Level of evidence**
We cannot recommend either physiotherapy (including stretching, soft-tissue work) or no physiotherapy for individuals with foot issues with Friedreich ataxia.	—	⊕◯◯◯
Justification: There is insufficient evidence to support the use of physiotherapy for foot issues with Friedreich ataxia.		
Subgroup considerations: This recommendation is for individuals with Friedreich ataxia with foot issues.		

Lay summary

Lay summary of clinical recommendations for foot deformity in Friedreich ataxia

Why these recommendations?

Individuals with Friedreich ataxia can have foot problems that make it difficult to stand, walk or transfer. There are some treatments for foot problems that might help to improve or maintain mobility.

There have not been many studies of the effect of surgery for foot issues in Friedreich ataxia. However, the opinion of healthcare experts who work with people with Friedreich ataxia is that surgery may be considered if it would benefit foot alignment which could help with walking and/or safety and independence in transfers.

Before considering surgery, it is important that alternatives such as ankle-foot orthotics (AFOs) have been tried. AFOs can help to stabilize the ankle when walking, prevent foot deformity, improve posture and standing, and improve walking. In particular, early and regular use of AFOs

may mean a person with Friedreich ataxia and foot issues may not need surgery to correct foot deformity.

What does this mean for you as a person living with Friedreich ataxia or caring for someone living with Friedreich ataxia?

It might be important for you to speak with your healthcare provider about Friedreich ataxia and foot deformity and what it means for you. When your healthcare provider is talking to you about the option of foot surgery and how it might help, it is important to also consider the risks before going ahead. These include the risk of infection, pain associated with surgery, possible loss of mobility in the recovery phase after surgery, managing casts/immobilization if that is needed, and the risk of complications during surgery (e.g., bleeding, heart issues).

Who are these recommendations specifically for?

These recommendations are specifically for individuals with Friedreich ataxia with foot deformity.

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