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Case Report

Intensive Multimodal Treatment for A Young Adult with Friedreich Ataxia: A Case Report

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ABSTRACT

Background: Friedreich's ataxia (FRDA) is a progressive, neurodegenerative autosomal recessive disorder affecting multiple systems of the body. Physical therapy has been found to be beneficial for improving function and quality of life in individuals with FRDA. However, there is little evidence supporting specific interventions that would address functional concerns in these patients, and the most optimal rehabilitation strategy has yet to be determined.

Case Presentation: The participant was a twenty-six-year-old female who had been diagnosed with FRDA ten years earlier. After discontinuing physical therapy for three years, she started using a wheelchair and her function declined. The primary outcome measures employed to assess FRDA were the scale for assessment and rating of ataxia (SARA) and the functional independence measure (FIM). Other measures included strength, range of motion and neurologic screening. At initial examination, the participant presented with muscle weakness, poor balance and coordination, and mobility restrictions. The patient received intensive physical therapy with five weekly sessions over a period of three months. Interventions focused on coordinative, balance, and strength training activities.

Results: The patient demonstrated a 30-point increase in the FIM, a 9-point decrease in the SARA, and amelioration in manual muscle testing scores upon completion of the rehabilitation program.

Conclusion: The FRDA patient displayed improvement in all outcome measures. Strength, ataxia severity and functional abilities were enhanced while a higher level of independence was gained. Our observations suggest that an intensive multimodal approach holds potential in the management of FRDA and call for further research.

KEYWORDS: Friedreich's ataxia, Rehabilitation outcomes, Physical therapy

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INTRODUCTION

Friedreich's ataxia (FRDA) is an autosomal recessive neurodegenerative disease that affects multiple systems of the body [1]. Although rare, it is the most common form of hereditary ataxia in Europe, the Middle East and the United States [1]. The estimated prevalence of FRDA is approximately 1 in 50 000

people, affecting males and females equally [1]. This condition is caused by a mutation in the frataxin gene in chromosome 9, which may alter the mitochondrial iron homeostasis [1].

FRDA symptoms typically manifests during puberty and the most prominent clinical features include progressive ataxia, loss of joint position sense and pyramidal muscle weakness [2]. Over the course of the disease, patients gradually develop mobility impairments and wheelchair dependency occurs within a decade from symptom onset [2]. Apart from neurological manifestations, individuals with FRDA might develop cardiac, endocrine and musculoskeletal pathologies such as hypertrophic cardiomyopathy, diabetes mellitus, scoliosis, and pes cavus [2].

On average, the mean life expectancy in this population is reported to be 36.5 years with the leading cause of mortality being cardiac dysfunction [2].

At present, there is no effective pharmacological treatment or cure for FRDA [3]. Intervention efforts are focusing on rehabilitation strategies to address both neurological symptoms and secondary musculoskeletal deficits in this disease [3]. Accordingly, recent studies found that adherence to a rehabilitation program is beneficial for recovery during acute events and for amelioration of long-term outcomes in FRDA [3]. However, there is minimal evidence supporting specific physical therapy procedures that would manage the disease's deleterious impact on mobility and function [3].

The literature suggests using treatment strategies similar to those in other degenerative neurological conditions [4]. Hence, primarily focusing on teaching strategies and compensatory techniques that aim to maintain or improve a person's ability to participate at the level one wishes in one's environment and role in the community [4]. Conventional physical therapy interventions may include balance and coordination programs, aerobic conditioning, task-orientated functional training, stretching, strength exercises, and gait training using adaptive equipment [4].

There is emerging evidence suggesting that balance and trunk-limb coordination training enhances ataxia and motor performance in adults with degenerative cerebellar ataxia including FRDA [5]. In addition, there is data indicating that interventions consisting of strength and physical conditioning provided to individuals with FRDA showed significant clinical improvement in their function [6].

Few studies employed a multi-faceted program focusing on therapeutic strengthening exercises, as well as on coordinative and balance training [7]. Miyai et al. (2012) reported that a rehabilitation program including intensive strength, balance and coordinative practice improved ataxia, function and number of falls in individuals with spinocerebellar and idiopathic cerebellar ataxia [8]. In addition, Nardone et al. (2014) demonstrated that interventions targeting static and dynamic balance control, gait training, flexibility and strengthening significantly improved balance and cadence during gait in participants with degenerative cerebellar ataxia [9]. However, no studies have directly investigated the potential rehabilitative role of such interventions combined in improving motor performance and in reducing ataxic symptoms in the FRDA population.

Given the paucity of data specifically describing the interventions and their impact on FRDA symptoms, the purpose of this case study is to report the outcomes of a multimodal treatment approach consisting of strength, coordination, and balance training on ataxia severity and function for a young adult with FRDA over a three-month period.

CASE PRESENTATION

Patient Description:

The patient is a twenty-six-year-old female who was genetically diagnosed at sixteen years of age with FRDA. Her medical history indicated that she had no cardiomyopathies nor any endocrine pathologies. She did not take any medications, except for an antioxidant supplement Coenzyme Q10. The last time the patient participated in a physical therapy intervention was about three years ago in 2019.

After she discontinued her maintenance

program due to the covid-19 pandemic, general deconditioning was reported. This resulted from the lack of activity and the decreased adherence to the prescribed home exercise program. Shortly after, she transitioned to using a wheelchair after sustaining many falls and injuring herself owing to the progression of ataxia. The patient namely suffered from a fifth metatarsal hairline fracture in the left foot and a vertical stable right patellar fracture. Both injuries further influenced the patient's ability to walk as well as her ambulatory confidence. Sub sequently, she decided not to use an assistive device for ambulation but to move straight to wheelchair use, considering that the latter was a safer mobility mean and not as physically and mentally exhausting as assistive devices.

Moreover, the patient reported a loss of coordination in her hands as she faced progressive difficulties in performing daily activities such as eating, drinking, and writing. She also stated encountering some environmental barriers at her home, which further restricted her ability to transfer and carry out self-care activities independently. Currently, the patient is unemployed and unwed, living with her parents in a second floor apartment with an elevator access. Apart from that, the patient has a strong social support network including her family members and friends.

The patient was referred for physical therapy from her physician to address the muscle mass loss and the declining functional status. Upon evaluation, the patient's primary rehabilitation goals were to regain strength and improve her independence. The subject gave a written informed consent in accordance with the Declaration of Helsinki prior to the participation in the study.

Examination Procedures:

A series of measures were performed at baseline and immediately after the treatment. The testing procedures involved executing a detailed musculoskeletal and neurological examination to help establish the extent of neuropathy and motor performance ability of the FRDA subject [10].

Range of motion and strength were tested using goniometry and manual muscle testing respectively [11]. Upper extremity assessment revealed full range of motion in all joints. In the lower extremity, ankle dorsiflexion and foot eversion were both restricted bilaterally. Pes cavus was detected as both feet had elevated medial arches, forefoot valgus, hindfoot varus, reduced subtalar pronation and a shortened gastrocnemius and Achilles tendon.

The trunk and upper extremity muscles were mildly weak. However, in the lower extremity significant weakness and muscular atrophy were detected proximally, namely in hip extensor and abductor muscles. Simultaneously, distal weakness was noted mostly in ankle dorsiflexor and evertor muscles. The right lower limb was slightly weaker than the other side, which might have been related to the prior right patellar fracture. Screening for scoliosis was done as it is a common feature of FRDA, nevertheless the patient did not have such condition [11].

The sensory integrity was evaluated [11]. Joint position sense and light touch were mildly attenuated proximally to become diminished distally throughout the extremities in a symmetrical distribution. Pain and temperature sense were intact in both upper and lower extremities. The patient did not experience sensations of neuropathic pain such as burning or electric shock-like pain neither pins and needles sensation. Furthermore, cranial nerves were evaluated and revealed no evidence of impairment in the ocular movements and visual field, nor demonstrated the presence of visual acuity deficits, hearing loss, dysarthria, and dysphagia.

Reflex integrity was examined, with an emphasis on deep tendon reflexes [11]. In the lower extremity the deep tendon reflexes were absent (quadriceps and gastrocnemius 0/4), and in the upper extremity they were decreased bilaterally (biceps and triceps 1/4 and brachio-radialis 2/4). A positive Babinski reflex was apparent indicating corticospinal tract involvement. Muscle tone testing did not show any signs of spasticity or involuntary painful muscle spasm.

Specific Scales and Measures:

The evaluation procedures also involved functional measurement tools. The scale for the assessment and rating of ataxia (SARA) and

the functional independence measure (FIM) were the main outcome measures used to evaluate balance, coordination, mobility and activities of daily living [18].

The scale for the assessment and rating of ataxia (SARA) is one of the most commonly employed tools for assessing FRDA, demonstrated to have good reliability and validity [12]. The SARA score is designed to evaluate cerebellar symptoms, and to measure the severity of ataxia [12]. Thus, it could be used to evaluate the progression of the disease as well as the efficacy of the rehabilitation program [12]. The scale has a score ranging from 0 (no ataxia) to 40 (most severe ataxia) [12].

It is made up of eight items each with specific numeric scores related to stance, sitting, speech, gait, finger-chase test, nose to finger test, heel-shin test and fast alternating movements [12].

The patient had a SARA score of 28⁄40 reflecting a moderate severity of ataxia. During assessment, the patient demonstrated a poor standing dynamic balance. She was able to stand in a natural position only with a strong support, and her stance was marked by an increase in postural sway amplitude. Whereas, assuming a standing position with a narrow base of support was impossible. Likewise, static sitting examination denoted poor balance as the patient relied on her upper extremity for stability. In addition, no speech disturbances were apparent.

In observational gait analysis, the patient managed to walk with the support of two parallel bars. Nevertheless, she was incapable of completing the 10-meter pre-set trajectory, covering only a distance of 4.5 meters. Asymmetrical and variable feet placement led to an irregular and unstable stumbling path. Accordingly, the patient demonstrated an ataxic gait pattern that resulted in an impaired central displacement amplitude, directional inaccuracy and an impaired control of the movement's force. In this case, longer gait cycles were observed, in which a shorter step length and a greater step width were evident besides an overall decrease in gait speed. Further, a significant increase in double stance duration was noted; the transfer of weight from leg to leg was very slow in order to compensate for the wide oscillation of the centre of mass and it is indicative of poor dynamic balance and stability. The patient also displayed a reduced swing phase time, a slower swing velocity and a greater stance phase duration. The aforementioned reflected the patient's reduced ability to maintain balance whilst in single leg stance which is a highly unstable body configuration. Concurrently, the patient also demonstrated a steppage gait pattern. An increase in hip and knee flexion accompanied by toes hyperextension were observable during the swing phase. The latter is a well-established motor strategy aimed at compensating for the reduced dorsiflexion and the weak tibialis anterior in order to promote foot clearance.

Moreover, decreased coordination in all limbs was evident. In the finger chase and finger-nose manoeuvres, dysmetria accompanied by a mild intentional tremor indicated fine motor skills deterioration (more tremor noted on the left side). In addition, the patient could not perform the heel-shin slide test for she was not able to perceive the position of her joints while moving her leg in order to accomplish the task. Lastly, dysdiadochokinesia was demonstrated during the fast alternating hand movement test. Impairment was seen with irregularity of the slowed down rhythm and amplitude of arm pronation and supination.

The functional independence measure (FIM) is a comprehensive functional test commonly used in FRDA studies [12]. It has been established as a valid and reliable tool in assessing the level of independence in functional activities [13]. This tool is an 18-item, seven-level, ordinal scale intended to be sensitive to changes over the course of a comprehensive rehabilitation program [13]. It comprises several measures of independence for self-care including sphincter control, transfers, locomotion, communication, and social cognition [13]. These components are graded according to the level of assistance an individual need; they range from total independence to total assistance [13]. FIM could also complement other ataxia specific tools in

assessing overtime changes in the patient's condition, while demonstrating that a better mobility correlates with a greater quality of life [12].

The total FIM score in this case was 86/126 reflecting activity limitations and participation restrictions. The patient was dependent in all transfer activities, and she faced difficulty in performing self-care functional tasks such as dressing, bathing and grooming. However, the patient's cognitive and communication functions were well preserved.

Physiotherapy Diagnosis and Prognosis:

The patient had an impaired motor function and sensory integrity associated with progressive disorders of the central nervous system [14]. Clinical findings included prominent muscle weakness, amyotrophy, areflexia and proprioceptive loss in the lower extremity, in addition to pes cavus and Babinski sign. Deterioration of gait, balance and coordination reflected a moderate severity of ataxia. They were coupled with limitations in daily activities, mainly locomotion and self-care tasks, besides participation restrictions which hindered engagement in social activities. The patient's main goal was to return to her normal activities at home and community, including being able to independently care for herself and transfer. The prognosis of the subject was good based on the mild clinical manifestations of the disease, absence of comorbidities (cardiac and endocrine pathologies), the patient's young age, motivation, and the support of her friends and family.

Interventions:

The patient was scheduled to receive individualized five physical therapy sessions per week, one hour each, over a three-month period [15]. One specialized physiotherapist employed an intensive multimodal treatment [15]. It included a combination of compensatory and restorative approaches, guided by the patient's previously described clinical presentation and context [15]. Sessions were divide each into three components: strength, balance and coordinative training [16]. Throughout the intervention period, the level of exercise difficulty was increased [16-17]. However, program progression depended on how well each exercise was performed and completed, as well as on the patient's level of fatigue and motivation [16-17]. Therefore, fatigue was monitored throughout the sessions as it guided the number of repetitions & progression of exercises [16-17]. Each exercise was executed in a controlled manner for two to three sets with an interval of 2-3 minutes of rest in between to avoid exhaustion [16-17].

Consecutively, the number of exercise repetitions was adjusted to the patient's capacity starting with 8-10 repetitions and when the task became easier progressing to 12-15 repetitions maximum [16-17]. The different types of exercises that have been executed are detailed in Table 1.

The strengthening program was founded on regaining and sustaining control of the muscles of the trunk and extremities. [15-17] Hence, exercises for general core stability and limbs retraining were chosen based on the muscles that were found to be weak during testing and were adjusted to the patient's capabilities [15-17]. Pelvic girdle and trunk muscles were mainly targeted in order to manage the patient's key areas of impairment and limitations to function. Strengthening exercises were executed in either sitting or lying positions, and the muscle groups trained were daily altered to prevent tiredness during the 30 minutes allotted for this component [15-17]. When the participant's muscle strength was insufficient to conduct anti-gravity activity, the physiotherapist assisted the movement [16-17]. Exercises were progressed by employing multi-joint muscle exercises, increasing the amount of repetitions, decreasing therapist-assistance, or by applying resistance using free weights, resistance bands, or therapist-applied resistance [16-17]. Similarly, sitting on a mat table with no upper extremity support and sitting on an air-filled disc or foam board made the exercises more challenging [16-17].

Stretching the tightened calf muscles was employed aiming to improve ankles range of movement [16-17].

The exercise program focused as well on static and dynamic balance activities rather than direct gait training [16-18]. Therefore, for a total of 15 minutes, exercises were carried out in both

Table 1: Summary of Intervention Exercises

Exercise Category		Description
Balance training	Seated, stable surface, trunk unsupported	Side-to-side weight transfer, Trunk bending, Pelvic tilt , Arm reaches , Foot circles, Toe touches
	Seated, unstable surface (Physioroll)	Gentle bounce, Trunk rotation (hands on or free), Marching (hands on or free), Side-to-side roll, Back-and-forth roll, Foot circles (hands on or free), Alternate hands and/or feet movements, Forward reach
	Standing on stable surface, feet apart progressing to feet together	Weight shift (lateral and anteroposterior), Arm raises/reaching movements, Trunk rotation, Alternating hip flexion, Up on toes, Up on heels, Knee bend, Single-limb stance, Standing with eyes closed
Coordination training	Stepping and stooping	Stepping (lateral and anteroposterior), Cross-step front, Tandem stepping, Step onto 8-inch step, Arm and contralateral leg lift, Walk on line or feet on either side from line, Frenkel's exercises
	Bed mobility	Lying bent knee rotations, Rolling on mat, Lying to sitting, and sit to stand practice
	Bimanual coordination	Throw the ball up in the air and catch it with same hand, Catch the ball from the therapist and throw it back, Throw a ball from the right hand to the left and back, Tie bows out of ribbons/tie shoelaces, Fasten buttons (shirt in lap), String beads, Practice opening and closing lids of bottles with alternating hands
	Hand-arm coordination	Pile building blocks, Pile little toy bricks, Pick up a pencil and roll between thumb and each finger, Turn over playing cards, Collect items in a small jar (marbles, paper clips, buttons), Practice writing, tracing shapes, coloring
	Drinking	Pour water from one cup into another, Move one hand to chin then back to table, Pour water into a big cup, Take cup and move it to chin then back to table
Strength training	Seated on stable surface progressing to unstable surface (air-filled disk)	Long arc quadriceps, Knee flexion, Seated march, Resisted plantarflexion, Ankle dorsiflexion, inversion and eversion, Alternating upper and lower extremity flexion, Resisted shoulder press, Resisted alternating biceps curls, Resisted alternating hammer curls, Resisted shoulder abduction, Resisted alternating shoulder flexion, Resisted elbow and shoulder extension kick-back
	Lying down	Quadruped position, Shift weight in quadruped position, Half cobra push up, Bridging, sit-ups, Clam/ hip abduction, isometric quads and glutes, Heel to chin, Heel slides to flex and extend the knee, Knees bent hip flexion and rotation, Cross body leg lifts, Ankle dorsiflexion, plantarflexion, inversion and eversion, Shoulder flexion and abduction, Elbow flexion and extension, Calf and hamstrings stretch

Adapted from Milne SC et al. [16] and Corben LA et al. [17].

seated and standing positions with the goal of enhancing postural control [16-18]. They started with stabilizing in a challenging static position and progressed to dynamic arm and leg movements in the same or modified position [16-18]. When needed the physiotherapist provided stabilization "hand's on" [16-18]. The exercises were progressed by reducing the amount of upper extremity support used, from holding on to a stable object (e.g. walker, chair, parallel bars) with two hands, then with one hand, to no support [16-18]. Similarly, different surfaces such as mat table, foam board, or balance disc were used to increase the difficulty of the seated exercises [16-17].

Lastly, coordinative training was administered for 15 minutes [5, 16]. It involved the practice of synchronous functional tasks using guided movement with progressively less assistance [5, 16].Exercises targeted fine and gross motor skills, and eye-hand coordination [5, 16]. They included functional bed mobility, stepping exercises, bimanual coordination exercises, and hand-arm coordination exercises [5, 16]. Activities were broken down into components that are simple enough to be performed correctly with a slow speed [5, 16]. Assistance was provided when necessary with the use of sensory cues (tactile, visual, and proprioceptive) [5, 16].

In addition, strategies such as decreasing the degrees of freedom and speed (through behavioural means or increasing limb inertia) during serial movements have been offered as an accommodative strategy to further help improve motor performance [5, 16]. Exercises were progressed through reduction of cues from therapist and by increasing the range and speed of the movement [5, 16]. They were also advanced form single-joint to multi-joint movements [5, 16].

Apart from that, the patient's environment was optimized [17]. Therapeutic adaptive equipment was provided to support function and

that was valuable for teaching practical everyday strategies aiming to promote independence in daily activities [17]. Hence, few home modifications were implemented like installing bath tub grab bars, safety handles, and a toilet safety frame to support independent toileting [17]. Similarly, bed support rails were installed to assist in repositioning while in bed and assist in transitioning in and out of bed [17]. In addition, stand aids and high sitting sofas were placed alongside furniture risers for arm chairs in order to offer support during sit to stand transfers [17].

RESULTS

At the completion of the three-month rehabilitation program, the patient was reassessed and the outcome measures were encouraging. The participant perceived improvements in balance, strength and coordination alongside a higher level of autonomy.

Upon re-evaluation, no changes were detected in range of motion nor in sensory and reflex functions, whereas manual muscle testing scores ameliorated, noticeably in the upper extremity (Appendix 1). Additionally, the participant's SARA score decreased 9 points (Appendix 2) representing an improvement in ataxia. Coordination was refined mostly in the upper limbs for they displayed a reduction in dysmetria and intentional tremor, in conjunction with revealing a more precise and rhythmic alternating hand movements. No marked observational changes in the gait pattern were perceived. However, the patient gained an ability to walk with a strong support for a longer distance of approximately 12 meters, as well as to sit and stand unsupported which in turn indicated an advancement in balance and strength.

A 30 points increase in FIM score reflected that the functional limitations found from the initial evaluation improved, mainly safety and independence during functional activities (Appendix 3). The patient continues to require minimal assistance to dress the lower body, and moderate assistance to negotiate stairs. Yet, she was independent in all other tasks and had the capacity to perform with no help transfers, self-care, and locomotion activities. Further, the participant reported that she felt stronger, more confident and steadier with activities of daily living performance.

DISCUSSION

The purpose of this case report was to investigate the outcome of an intensive 3-month multimodal physical therapy treatment approach consisting of strength, balance and coordination training for a patient with FRDA. Our patient's neuromuscular integrity and motor performance were assessed before and after the intervention period using both traditional sensorimotor evaluation techniques and functional measurement tools SARA and FIM. The participant's main goal at the initial examination was to regain autonomy and strength in daily activities. Following the intervention period, tests scores indicated amelioration in all outcome measures when compared to baseline scores. Improvements were reported in balance, strength, coordination, mobility and performance of activities of daily living.

In this regard, the participant's SARA score decreased from 28/40 to 19/40 with the intervention. Consequently, multi-joint coordination and balance improved, and while the patient revealed no significant difference in her gait pattern, her gait ability however was ameliorated. The functional gains were further confirmed by the patient's subjective reports of experiencing increased control during upper extremity movements mainly in handling utensils.

The following results might be explained by the fact that treatments focusing on coordinative and balance training may reduce ataxic symptoms and improve motor performance, which in turn contributes to a greater gait function for patients with cerebellar ataxia [5, 18]. The observed results were consistent with previous studies. Correspondingly, several systematic reviews recommended intensive physical therapy for cerebellar ataxia, which focused on balance and muscle strengthening [4, 15]. They suggested that such interventions could improve the SARA score [4, 15].

Likewise, other studies showed that intensive coordinative training in individuals with

degenerative cerebellar disease, significantly improved the SARA score, ataxia symptoms and gait ability [5, 8].

Concomitantly, evidence indicated that strength training, predominantly core stability exercises coupled with dynamic task practice seemed to be an important intervention for people with cerebellar dysfunction to improve gait and balance scores, as well as to facilitate skilled motor behaviour of the upper extremities [15].

Furthermore, it is important to note in the following case that stability was seen in sensory and reflex integrity, also minimal changes were apparent in lower extremity coordination. Nevertheless, the previously mentioned may not be negative findings given the chronic and progressive nature of FRDA [10].

The patient scored 86/126 on the FIM at initial examination and 116/126 post-treatment. The results reflected an increase in the patient's levels of independence and function, suggesting that the adopted interventions may be beneficial for mobility and motor function in people with FRDA.

Small studies with similar results provided some evidence in support of implementing strengthening exercises in conjunction with balance and coordination retraining for individuals with ataxia [14, 16]. It was found that the aforementioned were able to improve motor performance in daily activities and reduce limitations while supporting increased participation [14, 16]. It was also reported that Individuals with FRDA who were dependent on a wheelchair may still benefit from rehabilitation to improve their mobility and transfer ability [4, 16].

Accordingly, additional studies highlighted the importance of adequate sitting balance, trunk control and coordination for improving mobility measures in people with afferent ataxia including walking, standing, transferring and upper limb function [9, 18].

Moreover, the augmentation of the FIM score might be as well a consequence of the environmental modifications invoked. For instance, the literature documented that a compensatory approach, which includes movement retraining, assistive devices and optimizing the environment, seems valuable for enhancing independence in individuals with FRDA, not to mention beneficial for teaching practical strategies of managing the condition [4, 17].

Apart from that, extremities and core strength gains were demonstrated through increased manual muscle testing scores and through progression in ambulation, sitting and standing capacity. The visible improvements might be directly attributed to the patient's plan of care. Hence, the beneficial aspects of a multimodal rehabilitation program can be explained theoretically by the fact that therapeutic exercises may induce morphological and physiological changes within the contractile tissues, which in turn results in muscle fibres hypertrophy and enhanced motor unit recruitment [6, 19].

On that account, previous reports recommended employing an overall treatment that focuses on maintaining joint integrity and general muscle conditioning in individuals with ataxia. Concurrently, further trials revealed that such interventions might reverse or halt the downward decline in function seen in most people with FRDA [6, 19].

Another reason for the favourable functional outcomes might be related to the intensity of rehabilitation. Few investigations argued that higher training intensities were associated with greater improvements in clinical outcomes [16, 20]. They suggested that the rehabilitation duration in FRDA should be at least 4 weeks including more than 1 hour of training for 3 days per week [16, 20].

Finally, we found a limited number of studies with high methodological quality and scientific evidence inquiring into rehabilitation outcomes for patients with FRDA [19-20].

Hence, discussion of our findings was constrained. Given the limited evidence on treatment of patients with FRDA, additional studies are needed to determine the optimal duration, intensity, and specific interventions for this population.

CONCLUSION

In summary, a three-month period of intensive rehabilitation focusing on strength, balance and coordination training demonstrated positive gains in all outcome measures in the FRDA patient. As a result, the participant perceived improvements in strength, ataxia severity and functional abilities that were coupled with a higher level of independence.

Nonetheless, general inferences cannot be drawn from a case report. Therefore, it is thought that further studies with a larger sample of participants will be required in order to verify the effectiveness of the interventions utilized in this case report for other individuals with FRDA. Likewise, it is equally important to investigate the long-term outcomes of such interventions through a six-month to 1-year follow-up.

Authors' contributions

Grace Battal: Designed the study and collected the data**;** Analyzed the data and wrote the manuscript.
Nicolas Pinsault: Contributed to data analysis and manuscript revision
Berthe Hanna-Boutros: Supervised the study and revised the manuscript

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Appendix 1: Manual Muscle Testing Scores.

Muscle groups		**Admission**		**Discharge**	
Lower extremity		**Right**	**Left**	**Right**	**Left**
Hip	Flexors	3-	3-	3+	3+
	Extensors	2-	2	3-	3
	Abductors	2-	2	3	3
	Adductors	3-	3-	3-	3-
Knee	Flexors	2+	3-	3	3
	Extensors	2+	3-	3+	3+
Ankle	Plantarflexors	3+	3+	4-	4-
	Dorsiflexors	2-	2-	3-	3-
Foot	Invertors	3-	3-	3	3
	Evertors	2-	2-	3-	3-
Upper extremity		**Right**	**Left**	**Right**	**Left**
Shoulder	Flexors	3+	3+	4+	4+
	Extensors	3+	3+	4+	4+
	Abductors	3+	3+	4+	4+
	Adductors	3+	3+	4+	4+
Elbow	Flexors	4-	4-	4+	4+
	Extensors	4-	4-	4+	4+
Wrist	Flexors	4-	4-	4	4
	Extensors	4-	4-	4	4
Trunk					
Trunk	Flexors	2		3	
	Extensors	2		3	

Grading Scale Range: 0 to 5. 0: No visible or palpable contraction. 1: Visible or palpable contraction (no range of motion). 2- : Partial range of motion, gravity eliminated. 2: Full range of motion, gravity eliminated. 2+: Gravity eliminated/slight resistance or less than range against gravity. 3-: more than half but not full range of motion, against gravity. 3: Full range of motion against gravity. 3+: Full range of motion against gravity, slight resistance. 4-: Full range of motion against gravity, mild resistance. 4: Full range of motion against gravity, moderate resistance. 4+: Full range of motion against gravity, almost full resistance. 5: Normal, maximal resistance. Adapted from Nicola JP [11].

Appendix 2: Scale for Assessment and Rating of Ataxia (SARA).

<table>
<tr><td colspan="2">1) Gait</td><td colspan="2">2) Stance</td></tr>
<tr><td colspan="2">Patient is asked (1) to walk at a safe distance parallel to a wall including a half-turn (turn around to face the opposite direction of gait) and (2) to walk in tandem (heels to toes) without support.

0: Normal, no difficulties in walking, turning and walking tandem (up to one misstep allowed).
1: Slight difficulties, only visible when walking 10 consecutive steps in tandem.
2: Clearly abnormal, tandem walking, more than 10 steps not possible.
3: Considerable staggering, difficulties in half-turn, but without support.
4: Marked staggering, intermittent support of the wall required.
5: Severe staggering, permanent support of one stick or light support by one arm required.
6: Walking more than 10 meters only with strong support (two special sticks or stroller or accompanying person).
7: Walking less than 10 meters only with strong support (two special sticks or stroller or accompanying person).
8: Unable to walk, even supported.</td><td colspan="2">Patient is asked to stand (1) in natural position (2) with feet together in parallel (big toes touching each other) and (3) in tandem (both feet on one line, no space between heel and toe). Patient does not wear shoes, eyes are open. For each condition, three trials are allowed. Best trial is rated.

0: Normal, able to stand in tandem for more than 10 seconds.
1: Able to stand with feet together without sway, but not in tandem for more than 10 seconds.
2: Able to stand with feet together for more than 10 seconds, but only with sway.
3: Able to stand for more than 10 seconds without support in natural position, but not with feet together.
4: Able to stand for more than 10 seconds in natural position only with intermittent support.
5: Able to stand for more than 10 seconds in natural position only with constant support of one arm.
6: Unable to stand for more than 10 seconds even with constant support of one arm.</td></tr>
<tr><td>Score pre-treatment</td><td>7</td><td>Score pre-treatment</td><td>6</td></tr>
<tr><td>Score post-treatment</td><td>6</td><td>Score post-treatment</td><td>3</td></tr>
<tr><td colspan="2">3) Sitting</td><td colspan="2">4) Speech disturbance</td></tr>
<tr><td colspan="2">Patient is asked to sit on an examination bed without support of feet, eyes open and arms outstretched to the front.

0: Normal, no difficulties sitting for more than 10 seconds.
1: Slight difficulties, intermittent sway.
2: Constant sway, but able to sit for more than 10 seconds without support.
3: Able to sit for more than 10 seconds, only with intermittent support.
4: Unable to sit for more than 10 seconds</td><td colspan="2">Speech is assessed during normal conversation.

0: Normal.
1: Suggestion of speech disturbance.
2: Impaired speech, but easy to understand.
3: Occasional words difficult to understand.
4: Many words difficult to understand.
5: Only single words understandable.
6: Speech unintelligible / anarthria.</td></tr>
</table>

without continuous support.			
Score pre-treatment	4	**Score pre-treatment**	0
Score post-treatment	2	**Score post-treatment**	0

Adapted from Saute JA et al. [12].

<table>
<tr><td colspan="3">5) Finger chase</td><td colspan="3">6) Nose-finger test</td></tr>
<tr><td colspan="3">Rated separately for each side.
Patient sits comfortably. If necessary, support of feet and trunk is allowed. Examiner sits in front of patient and performs 5 consecutive sudden and fast pointing movements in unpredictable directions in a frontal plane, at about 50 % of patient´s reach. Movements have an amplitude of 30 cm and a frequency of 1 movement every 2 seconds. Patient is asked to follow the movements with his index finger, as fast and precisely as possible. Average performance of last 3 movements is rated.
0: No dysmetria.
1: Dysmetria, undershooting or overshooting target less than 5 centimeters.
2: Dysmetria, undershooting or overshooting target less than 15 centimeters.
3: Dysmetria, undershooting or overshooting target more than 15 centimeters.
4: Unable to perform 5 pointing movements.</td><td colspan="3">Rated separately for each side.
Patient sits comfortably. If necessary, support of feet and trunk is allowed. Patient is asked to point repeatedly with his index finger from his nose to examiner’s finger which is in front of the patient at about 90 % of his reach. Movements are performed at moderate speed. Average performance of movements is rated according to the amplitude of the kinetic tremor.
0: No tremor.
1: Tremor with an amplitude less than 2 centimeters.
2: Tremor with an amplitude less than 5 centimeters.
3: Tremor with an amplitude more than 5 centimeters.
4: Unable to perform 5 pointing movements.</td></tr>
<tr><td>Score pre-treatment</td><td>Right side: 2</td><td>Left side: 3</td><td>Score pre-treatment</td><td>Right side: 2</td><td>Left side: 3</td></tr>
<tr><td>Score post-treatment</td><td>Right side: 1</td><td>Left side: 2</td><td>Score post-treatment</td><td>Right side: 1</td><td>Left side: 2</td></tr>
<tr><td>Mean of both sides pre-treatment (Right + Left)/2</td><td colspan="2">2.5</td><td>Mean of both sides pre-treatment (Right + Left)/2</td><td colspan="2">2.5</td></tr>
<tr><td>Mean of both sides post-treatment (Right + Left)/2</td><td colspan="2">1.5</td><td>Mean of both sides post-treatment (Right + Left)/2</td><td colspan="2">1.5</td></tr>
<tr><td colspan="3">7) Fast alternating hand movements</td><td colspan="3">8) Heel-shin slide</td></tr>
<tr><td colspan="3">Rated separately for each side.
Patient sits comfortably. If necessary, support of feet and trunk is allowed. Patient is asked to perform 10 cycles of repetitive alternation of pronation and supination of the hand on his/her thigh as fast and as precise as possible. Movement is demonstrated by examiner at a speed of approximately 10 cycles within 7 seconds. Exact times for movement execution have to be taken.
0: Normal, no irregularities, performs in less than 10 seconds.
1: Slightly irregular, performs in less than 10 seconds.
2: Clearly irregular, single movements difficult to distinguish or relevant interruptions, but performs in less than 10 seconds.
3: Very irregular, single movements difficult to distinguish or relevant interruptions performs in more than 10 seconds.</td><td colspan="3">Rated separately for each side.
Patient lies on examination bed, without sight of his legs. Patient is asked to lift one leg, point with the heel to the opposite knee, slide down along the shin to the ankle, and lay the leg back on the examination bed. The task is performed 3 times. Slide-down movements should be performed within 1 second. If patient slides down without contact to shin in all three trials, rate 4.
0: Normal.
1: Slightly abnormal, contact to shin maintained.
2: Clearly abnormal, goes off shin up to 3 times during 3 cycles.
3: Severely abnormal, goes off shin 4 or more times during 3 cycles.
4: Unable to perform the task.</td></tr>
</table>

4: Unable to complete 10 cycles.					
Score pre-treatment	Right side: 2	Left side: 2	**Score pre-treatment**	Right side: 4	Left side: 4
Score post-treatment	Right side: 1	Left side: 1	**Score post-treatment**	Right side: 4	Left side: 4
Mean of both sides pre-treatment (Right + Left)/2	2		**Mean of both sides pre-treatment** (Right + Left)/2	4	
Mean of both sides Post-treatment (Right + Left)/2	1		**Mean of both sides post-treatment** (Right + Left)/2	4	
Total Score pre-treatment	**28/40**				
Total Score post-treatment	**19/40**				

Adapted from Saute JA et al. [12].

Appendix 3: Functional Independence Measure (FIM).

LEVELS			
Independent (no helper)		7: Complete Independence (Timely, Safely)	
		6: Modified Independence (Device)	
Modified Dependence (helper)		5: Supervision (Subject = 100%+)	
		4: Minimal Assist (Subject = 75%+)	
		3: Moderate Assist (Subject = 50%+)	
Complete Dependence (helper)		2: Maximal Assist (Subject = 25%+)	
		1: Total Assist (Subject = less than 25%)	
ITEMS		**ADMISSION**	**DISCHARGE**
Self-Care	A. Eating	**7**	**7**
	B. Grooming	**4**	**7**
	C. Bathing	**2**	**7**
	D. Dressing - Upper Body	**4**	**7**
	E. Dressing - Lower Body	**3**	**4**
	F. Toileting	**5**	**7**
Sphincter Control	G. Bladder Management	**7**	**7**
	H. Bowel Management	**7**	**7**
Transfers	I. Bed, Chair, Wheelchair	**2**	**6**
	J. Toilet	**2**	**6**
	K. Tub, Shower	**2**	**6**
Locomotion	L. Walk/Wheelchair (W walk, C wheelchair, B both)	**5 (C)**	**7 (C)**
	M. Stairs	**1**	**3**
Motor Subtotal Score		*51/91*	*81/91*
Communication	N. Comprehension (A auditory, V visual, B both)	**7 (B)**	**7 (B)**
	O. Expression (V vocal, N non-vocal, B both)	**7 (B)**	**7 (B)**
Social Cognition	P. Social Interaction	**7**	**7**
	Q. Problem Solving	**7**	**7**
	R. Memory	**7**	**7**
Cognitive Subtotal Score		*35/35*	*35/35*
Total FIM Score		**86/126**	**116/126**

Adapted from Mackintosh S. [13].