CASE REPORT

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Efficiency of a 3-week multicomponent rehabilitation on improving the function in a patient with Friedreich's ataxia – a case report

Delotvornost tronedeljne višekomponentne rehabilitacije na poboljšanje stanja kod bolesnika sa Fridrajhovom ataksijom

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Abstract

Introduction. Friedreich's ataxia (FA) is an autosomal recessive neurodegenerative disease. Ataxia, as the cardinal symptom, affects the trunk, with swaying, imbalance, and falls, as well as the limbs, with increasing difficulty in activities of daily living. Physical therapy has been recognized as a means of managing physical symptoms and maximizing function in affected persons. To our knowledge, there are no studies that have evaluated the effectiveness of proprioceptive neuromuscular facilitation (PNF) stabilization techniques in the rehabilitation of patients with such a diagnosis. **Case report.** We present a 26-year-old female with FA who had severe truncal and limb ataxia, speech difficulty, and poor walking ability. During the three-week rehabilitation, an individually tailored physical therapy program based on PNF stabilization techniques was applied. The implemented rehabilitation program resulted in an overall functional improvement. The reduction in ataxia was registered according to the Scale for the Assessment and Rating of Ataxia (SARA). The Functional Independence Measure (FIM) instrument – a component of locomotion, revealed greater independence in walking. **Conclusion.** A rehabilitation program based on PNF stabilization techniques may reduce ataxia and improve walking ability in patients with FA.

Key words:
friedreich ataxia; physical and rehabilitation medicine; treatment outcome.

Apstrakt

Uvod. Fridrajhova ataksija (FA) je autozomno recesivna, neurodegenerativna bolest. Ataksija kao glavni simptom zahvata trup, uz njihanje, nestabilnost i padove, kao i ekstremitete, uz sve teže obavljanje aktivnosti svakodnevnog života. Fizikalna terapija se prepoznaje kao sredstvo za tretman fizičkih simptoma i postizanje maksimalne funkcije obolelih osoba. Prema našim saznanjima, ne postoje studije koje su ocenjivale korisnost tehnika stabilizacije proprioceptivne neuromišićne facilitacije (PNF) u rehabilitaciji bolesnika sa tom dijagnozom. **Prikaz bolesnika.** Prikazujemo ženu staru 26 godina sa FA koja je imala težak oblik ataksije trupa i ekstremiteta, teškoće u govoru i slabu sposobnost hodanja. U toku tronedeljne rehabilitacije primenjen je individualno prilagođen program fizikalne terapije, baziran na tehnikama stabilizacije PNF. Primenjeni rehabilitacioni program rezultirao je opštim funkcionalnim poboljšanjem. Smanjenje ataksije je registrovano Skalom za procenu i ocenu ataksije (*Scale for the Assessment and Rating of Ataxia* – SARA). Instrumentom za merenje funkcionalne nezavisnosti (*Functional Independence Measure* – FIM) – komponenta kretanja, pokazana je veća nezavisnost u hodu. **Zaključak.** Rehabilitacioni program zasnovan na tehnikama stabilizacije PNF može smanjiti ataksiju i poboljšati sposobnost hodanja kod obolelih od FA.

Ključne reči:
ataksija, fridrajhova; medicina, fizikalna i rehabilitacija; lečenje, ishod.

Introduction

Friedreich's ataxia (FA) is a hereditary disease with progressive neurological features named after the German physician Nikolaus Friedreich who first described it in 1863 [1]. The first symptoms usually appear in childhood or adolescence, and instability is most often reported as the first main symptom [2]. The prevalence of this rare disease ranges from one case in 20,000 in southwestern Europe, while in northern and eastern Europe, the prevalence is lower: one case in 250,000 [3]. Traditionally viewed as a neurodegenerative disease, FA patients also develop cardiomyopathy, scoliosis, diabetes mellitus, and other manifestations [4]. The main manifestations of FA include symptoms of afferent and cerebellar

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ataxia, central muscle weakness, speech disorder, swallowing problems, inability to maintain balance and proprioception, and vibration sensation disorders [5]. Walking disorders start early and manifest as unstable gait. Limb ataxia manifests as a lack of precise movement, slowness of movement, intentional tremors, and a coordination disorder. Basic activities of daily living (ADLs) become difficult due to ataxia. Usually, most patients are wheelchair-assisted by the third decade of life [3]. There is no approved disease-modifying therapy for the treatment of this disease [6]. Physical therapy is recommended to alleviate symptoms and improve motor function. The evidence published so far indicates that rehabilitation is effective in reducing ataxia and improving balance, mobility, and function [4, 7, 8]. A wide range of physical therapy interventions is effective in treating the symptoms and controlling the progression of this disease. In the rehabilitation of ataxia, static and dynamic balance exercises, gait exercises, coordination, and exercises to increase proprioceptive awareness are implemented [9]. Most studies have shown a reduction in ataxia symptoms due to the use of balance exercises [10, 11], but there is insufficient high-quality evidence to form clear recommendations and guidelines for clinical practice. We present the successfully performed multicomponent physical therapy intervention involving proprioceptive neuromuscular facilitation (PNF) stability exercises in patients with FA that could improve the clinical practice of such a rare disease.

Case report

We present a 26-year-old female complaining of generalized ataxia, unable to walk independently. FA was diagnosed when she was 21 (in 2016). The family history was negative. Symptoms gradually progressed to severe gait instability. A year after the diagnosis, she started using a walker. She did not report any further worsening of ataxia symptoms after the SARS-CoV-2 infection in November 2020. Mobilizing independently indoors, she reached for furniture to provide support when walking; she had several falls. She reported walking independently on the stairs in the house, holding onto the handrail with both hands. She was not socially active regularly due to a walking disability. Over the last year, symptoms progressed to serious gait instability: she became dependent on using a wheelchair for distance mobility. Carnosine supplementation was performed. She described a regular home exercise program that considered muscle strengthening exercise with 1 kg weighted cuff resistance and a few min of cycling on a stationary bike. Family support was present. In October 2021, she was admitted to the Institute for Physical Medicine, Rehabilitation, and Rheumatology “Dr. Simo Milošević” in Igalo, Montenegro for a regular three weeks of rehabilitation provided by the healthcare system once a year.

Assessment

On admission, an observation revealed kyphoscoliosis and hand muscle wasting. There were no signs of other significant muscle wasting, tingling, or paresthesia, and no abnormalities in hearing or vision. Manual muscle testing (MMT) was used for assessing general muscular strength [12, 13], and the score was 4/5. She had dysmetria, dysdiadochokinesia, and intentional tremor bilaterally. Many words were difficult to understand. Clinical evaluation was performed through the Scale for the Assessment and Rating of Ataxia (SARA). The use of the SARA scale is valuable for assessing ataxia and rehabilitation planning [14, 15]. SARA test scored 24/40. There was no evidence of increased muscle tone. Straight leg raise (SLR) test was positive, left 60°, right 50°. She was independent in the sit-to-stand movement. Independent sitting balance in supported sitting was present, with intermittent sway while she was sitting unsupported. In a wide stance, she stood independently for less than 10 sec. Romberg was positive. She was able to walk a short distance with the support of two persons or with a walker and the support of one person. The Functional Independence Measure (FIM) scale [16, 17] component of locomotion was rated 8/14. Overall, she did not require any assistance to perform basic ADLs: eating, dressing, and personal hygiene.

Intervention

We implemented a neurorehabilitation program aimed to improve ataxia, stability, and walking ability to prevent and slow complications of primary illness (Figures 1–4). The rehabilitation program considered the following exercise sessions: manual massage twice a day for 45 min and occupational therapy once a day for 15 min. During rehabilitation, an individually-tailored, problem-solving physiotherapy approach consisting of PNF stabilization, walking, and proprioceptive exercises was conducted (Table 1). To prevent and slow the sequelae of primary illness, isotonic muscle strengthening exercises with optimal manual resistance were applied.

Fig. 1 – Stabilizing reversal in side-lying in a patient with Friedreich’s ataxia.

Fig. 2 – Rhythmic stabilization of a patient with Friedreich's ataxia in prone elbows support.

Fig. 3 – A patient with Friedreich's ataxia: two-wheeled walker support. Controlled stepping.

Fig. 4 – Frenkel exercises over slide board in supine.

PNF therapy intervention was carried out following the philosophy of the method and principles of facilitation, such as resistance, approximation, stretching, movement patterns, manual, verbal, and visual contact, correct position, and the therapist's body position [18]. PNF techniques, such as stabilizing reversal and rhythmic stabilization, may be applied to promote trunk stability within the different postures that constitute the developmental sequence [19]. Generally, improvements in balance have been attributed to PNF stabilizing techniques [18, 20–22]. To promote stability, as balance disorder is one of the main manifestations of FA, we applied the PNF techniques stabilizing reversal and rhythmic stabilization in different positions: half-ring sitting, ring sitting, side-lying, side sitting with and without arms support, prone on elbows, quadruped, and sitting with and without foot support. With rhythmic stabilization, the subject tries to maintain position while the therapist provides perturbances without any movement, while in stabilizing reversal, small movement is allowed. To promote stability in kneeling and standing, approximation was given through both sides of the pelvis equally and directed downward. She practiced sitting steady without leg support for 30 sec with her arms outstretched forward. In a half-ring sitting position and sitting without foot support, we performed reaching out for objects from different angles.

Table 1

Physical therapy program[1]

Problem	Physical therapy interventions
General muscle strength and endurance	Abdominal curls, prone fly. Bridge with approximation over knees and with isometric hold. Isotonic strengthening exercises with manual resistance for upper extremity and lower extremity muscles, 1–2 series, 10 repetitions.
Postural control	To promote stability, we applied PNF techniques rhythmic stabilization and stabilizing reversal in side-lying (Figure 1), sitting with and without foot support, half-ring sitting, side sitting with and without hand support, and prone on elbows (Figure 2), and quadruped. To promote stability in kneeling and standing, approximation was given through both sides of the pelvis equally and directed downward. We practiced sitting steady without foot support for 30 sec with arms outstretched forward. In half-ring sitting and sitting without foot support, we performed reaching out for objects from different angles.
Walking	Walking with parallel bars, physiotherapist's support, and instructions, just a few meters at first. At the end of the second week, we started walking with two-wheeled walker support. Controlled stepping was practiced under visual control (Figure 3). We gradually extended the walking distance.
Proprioceptive awareness	Frenkel exercises over the slide board in a supine (Figure 4) and, in progression, in a sitting position. In sitting, she also placed her foot in marked spots on the floor. Finger chase exercise for the upper extremity.
Reduced flexibility	Hamstrings stretching, hold-relax PNF technique, and from the third week active stretching in sitting.

[1] Problem-oriented physiotherapy program based on proprioceptive neuromuscular facilitation (PNF) stabilization techniques also included Frenkel exercises, walking exercises, and strengthening exercises.

Table 2

Scale for the Assessment and Rating of Ataxia (SARA) and Functional Independence Measure (FIM) test rating, before and after rehabilitation[2]

Outcome measure	Before rehabilitation	After rehabilitation
SARA test score	24/40	20/40
Gait	7/8	6/8
Stance	4/6	3/6
Sitting	1/4	1/4
Speech disturbance	4/6	4/6
Finger chase	1/4	1/4
Nose-finger test	2/4	1/4
Fast alternating hand movements	2/4	2/4
Heel-shin slide	3/4	2/4
FIM test (locomotion) score	8/14	10/14
Walk	3/7	5/7
Stairs	5/7	5/7

[2] SARA score before and after rehabilitation showed a decrease in sub-items scores of gait, stance, and tremor amplitude with nose-finger test and heel-shin slide. FIM score before and after rehabilitation showed improvement in the sub-item score of the walk.

Outcomes

At the final assessment, the patient reported greater endurance with supported standing and walking. SLR test was positive: left 70°, right 65°. SARA test scored 20/40: examination showed a decrease in sub-items scores of gait, stance, and tremor amplitude with the nose-finger test and heel-shin slide (Table 2). She was able to walk with walker support and supervision. FIM scale rating of 10/14 revealed improvement in locomotion.

Discussion

FA is a progressive neurological disease with several different manifestations. The current treatment of that genetic disorder is directed toward all individual manifestations. Science is constantly searching for effective disease-modifying therapy, and meanwhile, rehabilitation remains the base of treatment that controls symptoms and slows the progression of the disease [6]. In various neurological disorders, including FA, intensive rehabilitation in the institution

has been effective in improving patient function [23]. Improvement in balance may occur after 3 weeks, and improvement in ataxia requires at least 4 weeks of rehabilitation. In addition, multicomponent inpatient rehabilitation has greater effects than home exercise programs alone [4]. The home exercise program cannot achieve or maintain the benefits of multi-segment rehabilitation [6]. That emphasizes the importance of inpatient rehabilitation. However, a three-week rehabilitation is just a "drop in the ocean" for patients with FA [24]. To achieve the maximum level of functioning, it would be helpful to provide inpatient rehabilitation more than once a year. There are very few patients with this diagnosis, and this would not significantly burden the healthcare system.

As part of rehabilitation, there are a large number of therapeutic interventions that differ in type, intensity, and duration. However, in a systematic review, Milne et al. [8] state that the evidence suggesting that rehabilitation improves function, mobility, balance, and ataxia is consistent. Physical therapy interventions enable the improvement of muscle flexibility and strength, postural control, and movement dexterity. By acting on the individual manifestations of this neurological disorder, physical therapy alleviates the effect of ataxia on patient function [3]. One of the main goals of neurological rehabilitation is to improve gait function [25]. Balance exercises and coordination exercises are especially useful for the functional recovery of the patient. There are no available data from high-quality randomized trials on the effectiveness of physical therapy interventions. Marquer et al. [26] stated that the intensity, duration, and content of the rehabilitation programs would have to be better defined.

During a three-week rehabilitation, we implemented an individually tailored, problem-oriented physiotherapy program based on PNF stabilization techniques. Outcome measures in our study showed improvement in overall functional performance. Such improvement may be due to specific stabilization techniques. Improved trunk stability had an influence on the reduction of upper limb tremors. Applied techniques resulted in better walking ability and diminished ataxia.

PNF stabilization concepts are effective in the treatment of postural control in stroke rehabilitation [18, 22]. As far as we know, there are no studies that have evaluated the effectiveness of PNF stabilization techniques in patients with FA, so the results of our research are worthy of attention. One case report limits the generalization of our findings. Our case report indicates the effectiveness of a three-week multicomponent rehabilitation, but long-term follow-up of the patient was lacking. The systematic review from 2017 emphasized that it is not clear how long the effects of rehabilitation are maintained [8]. Future research should consider monitoring the functional status of patients after discharge to gain insight into the duration of rehabilitation outcomes.

Although the conclusion is based only on one case, this paper can provide preliminary guidance for physiotherapists in treating individuals with FA.

Conclusion

The findings of this study provide evidence that an individually tailored rehabilitation program based on PNF stabilization techniques can reduce ataxia and improve functional status in patients with FA. The stabilization techniques of the PNF approach should be considered and included during the short-term multicomponent rehabilitation of these patients.

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