

Aquatic physical therapy in the treatment of a child with merosin-deficient congenital muscular dystrophy: case report

Cinthy Patrícia de Albuquerque Santos¹, Ricardo Cristian Hengles¹, Fábio Navarro Cyrillo², Fernanda Moraes Rocco³, Douglas Martins Braga¹

ABSTRACT

This case report describes an aquatic therapy program for a child with Merosin-Deficient Congenital Muscular Dystrophy. **Objective:** This study sought to investigate the effect of aquatic physical therapy on the speed and the rate of energy expenditure while moving around on a flat surface, in addition to the functional reach of the upper limbs suffering from the proximal weakness that affects these patients seeking greater independence. **Methods:** The Motor Function Measurement (MFM) and the Functional Reach Test (FRT) were used as assessment tools; the Energy Expenditure Index (EEI) was measured in seated locomotion, as well as the time spent, and muscle activation was measured via electromyography (EMG). The program lasted 12 weeks and included activities to improve mobility and agility as well as reaching from the seated position. **Results:** In the MFM the change in the score of the two dimensions (D2 and D3) that the treatment focused on was 6.8%. The functional reach improved by 16 centimeters (cm) and the amount of time moving while sitting decreased by 19 seconds (s). Energy expenditure decreased by 252.31 beats per minute (bpm). **Conclusion:** The aquatic physical therapy was effective for agility improvement in seated locomotion and upper limb functionality of a 6-year-old child with Merosin-Deficient Congenital Muscular Dystrophy.

Keywords: Hydrotherapy, Muscular Dystrophies, Electromyography, Rehabilitation

¹ Physiotherapist, Association for Assistance to Disabled Children/Central Unit - AACD.

² Masters in Physiotherapy, Professor at the Centro Universitário São Camilo.

³ Physiotherapist, Association for Assistance to Disabled Children/Central Unit - AACD.

Mailing address:

Associação de Assistência à Criança Deficiente
Cinthy Patrícia de Albuquerque Santos
Avenida Professor Ascendino Reis, 724
CEP 04027-000
São Paulo - SP, Brazil
E-mail: cinthyafisio2011@live.com

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INTRODUCTION

The term muscular dystrophy has been adopted for a number of different myopathic disorders that share a genetic nature with well-defined characteristics involving primary volunteer muscles tending toward incurable progressive muscle deterioration.¹

Within this group is Congenital Muscular Dystrophy (CMD) with recessive autosomal inheritance, characterized by altered musculature noticed at birth. Two distinct groups are known according to the presence or deficiency of the protein merosin, with the of protein-deficient group being the more severe phenotype.²

The clinical presentation includes hypotonia, atrophy, and stationary muscle weakness, or with minimal changes, and the presence of joint retractions with proximal regions most affected, diminished or nonexistent reflexes, and good cognition. Most individuals do not acquire a gait, but are able to remain seated and this is the main form of travel. The respiratory impairment is intense and this is closely related to the prognosis of the disease, as well as the spinal deformities.^{1,2} In addition to pharmacological treatment, these patients require a multidisciplinary team for rehabilitation. Physiotherapy is one of the resources present in this process and should be started early, as soon as it is diagnosed, even if this is in the first months of life. One can plan a program of long-term treatment and establish some goals such as: preventing the deformities caused by the disease, preserving the autonomy of the patient, avoiding pain and muscle fatigue, offering adjustments when necessary, and guiding the family about the disease and/or the rehabilitation process.²

Aquatic physiotherapy is part of the rehabilitation process of these patients with the distinction of providing a degree of freedom of movements that are more difficult to carry out on the ground. Some of the goals of therapy are to maintain the range of motion and muscle activation, to train respiratory skills, and improve physical conditioning, thereby maintaining greater functionality.³

Although the effectiveness of interventions with aquatic therapy on patients with neuromuscular diseases is evident today, studies on patients with CMD are scarce and limited. It is very difficult to find eligible patients with the same functional level and also to establish evaluation parameters suitable to their characteristics of progressive functional loss.

OBJECTIVE

To verify the effect of aquatic therapy on speed and the index of energy expenditure while traveling seated on a flat surface, and on the functional reach of the upper limbs, in addition to providing information and assisting therapists in drawing up a plan of care that includes aquatic therapy for patients with merosin-deficient CMD, striving for greater independence for these individuals.

METHODS

This work is a case study with an interventional clinical prospective, developed at the Aquatic Therapy Association of Assistance to Disabled Children (AACD), according to the ethical principles for research involving human beings, according to Resolution 466/12 of the National Health Council. It was approved by the Research Ethics Committee at the AACD, under Nº 17015913.2.0000.0085. The person responsible for the patient signed an informed consent form (ICF).

The child was 6 years old, male, and white, was born at full term weighing 3 kg, it was a natural childbirth with cephalic presentation. There were no pre-natal and/or perinatal complications, and no surgical procedure was performed; there are 3 siblings with no disabilities, and the parents are not consanguineous.

With regard to his motor development, he showed developmental delay and hypotonia since his earliest days of life, he sat with support at 1 year of age and without support with 2 years and 6 months, when he also began to speak. He has never crawled or walked. The family sought a pediatrician who requested a muscle biopsy examination to seek a diagnosis when they obtained the diagnosis of Merosin-Deficient CMD at 3 years of age.

The physical examination shows muscle atrophy and hypotonia with diffuse retractions in elbows, wrists, hips, knees, and ankles. The range of motion of the upper limbs is reduced due to grade 3 muscle weakness according to manual testing, grade 3 in musculature of the cervical spine, and 2 in the lower limbs.⁴ He presents hyperlordosis of the lumbar spine and shortening of the flexor muscles of both hips, estimated at 30 degrees in the Thomas test. He performs ample passive abduction of the hips, has contracture in bending the right knee 10 degrees, has no discrepancy in the

lower limbs, and the left foot presents equines' deformity.

The sensory and cognitive functions are preserved, but the deep reflexes are hypoactive. He presents no respiratory, cardiac, or associated digestive impairments. With respect to functionality, the child does not perform any postural change independently. He is semi-dependent for feeding and dependent for dressing and hygiene. When placed in a sitting posture, he can release upper limbs to perform some functions, but presents impairments in balance and protection reactions in this posture. He began transferring while seated slowly, this being his only form of locomotion.

Evaluation

All evaluations were implemented at the beginning and at the end of the program of aquatic therapy by a single investigator with experience in caring for patients with a diagnosis of neuromuscular diseases (NMD's).

The measure of motor function (MFM) was used first, which consists of three dimensions: the first (D1) is called standing and transfers (13 items); the second (D2) is axial and proximal function (12 items); and the third, (D3) distal motor function (7 items with 6 evaluating the upper limbs). This tool has been validated in the Portuguese language and allows the measurement of motor dysfunction in the diagnostic phase, follow-up, and the setting up of the therapeutic efficacy.⁵

The Functional Reach Test (FRT) was used to measure the functional reach of the upper limbs, but modified for a sitting posture. The child was placed in the sitting posture with support for the trunk and feet, with the shoulders at 90°, elbows extended, wrist in neutral position, prone and fingers flexed hands (closed). The tape measure was placed parallel to the ground at the height of the acromion. The patient was instructed to lean forward as far as possible without removing the buttocks from support. Three attempts were made and the highest value was marked.⁶

The energy expenditure Inex (EEI) was adapted for seated locomotion, because the literature only has a calculation for the gait. This was performed using the formula: exercising heart rate - resting heart rate/traveling speed in the seated position (m/s). The time was also measured in seconds to travel time seated (TTS) on a flat surface a distance of 2.0 meters.⁷

To analyze muscle recruitment, the instrument used was a four-channel Miotec® brand EMG sensor to evaluate the surface

electromyography using electrodes with Ag/AgCl hydrogel, a solid gel adhesive conductor, and Mark Kendall 200[®] electrodes; these were connected to a laptop containing the program Miograph[®] to record patterns of muscle activation and the response level in relation to the effort. The electrodes were positioned on the main muscles responsible for lateral inclination of lower trunk: the quadratus lumborum and abdominals, and were isolated from the water by using bioclusive dressing according to the recommendations of the SENIAM (Surface ElectroMyoGraphy for the Non-Invasive Assessment of Muscles); the signal was obtained during travel for a distance of 2.0 meters while sitting in the water.⁸

Intervention

The child did aquatic physiotherapy twice a week for 35 minutes per session over a period of 12 weeks. The therapeutic swimming pool was 5x10 meters (m) with a water temperature of 33°C. During the protocol period, the child continued doing physical therapy on dry ground and occupational therapy.

The functional goal established in aquatic physiotherapy was to improve his speed of movement on flat surfaces and so be less dependent on family, as seen in the International Classification of Functioning - ICF, based on the activity item and participation (code d4200 - transferring oneself while sitting).⁹

Each session consisted of a period of exercises for the trunk and for muscle activation of the lower abdominal and side flexors of the trunk. Then, the training for transferring while sitting was conducted, seeking to include functional as well as fun activities (Chart 1).

RESULTS

The scores pre and post-intervention in the dimensions of the MFM are reported in Table 1. The total score in dimensions 1, 2, and 3 presented significant improvement of 4.29%; in the individual scores, the highest percentage of improvement was observed in dimension 3 (10.33%), which is for distal movement. But considering the percentage of improvement of only the dimensions focused on in the study (2 and 3), this presents an overall score of 6.44%, since the patient has no abilities of orthostatism and gait, essential for the realization of the tests of dimension 1 - Standing and transfers.

The FRT saw some improvement, with 6 cm before and 22 cm after the treatment period (Table 2). The time it took for the child to travel a distance of 2.0 meters was also calculated for sitting on a flat surface. Before treatment, the TTS was 36 seconds, whereas after the intervention it was 15 seconds, reducing the time by 19 seconds for this transfer (Table 2).

With respect to the IEE, the patient obtained an initial index of 380 bpm and at the end of the period it went down to 107.69 bpm showing less energy expenditure during travel while seated.

In the analysis of the dynamic electromyography (EMG) of the quadratus lumborum and abdominal muscles, asymmetrical activations were observed before the intervention. The right quadratus lumborum muscle showed 120.1 μ v, the left 270.1 μ v, the right rectus abdominis 174.1 μ v, and left 272.1 μ v, demon-

strating a greater activation of the left hemibody. After the intervention, greater muscle recruitment was noted on the right, with the values of the right lumborum of 283.4 μ v, left lumborum of 263.5 μ v, right rectus abdominis of 335.7 μ v, and rectus abdominis side of 339.2 μ v, showing a more effective and symmetrical contraction in EMG parameters.

DISCUSSION

This case report describes a 12-week aquatic therapy program planned to improve muscle activation, agility, and especially the functionality during seated travel for a child of 6 years with merosin-deficient CMD. The results included improvement in motor function, improvement in travel time seated, as well as improvement of the reach of upper limbs in this posture, decrease in energy expenditure,

Chart 1. Summary of activities and aquatic exercises

Activities of stretching and muscle flexibility	Passive mobilization of hip and trunk; Passive stretching for hip flexors, knee extensors, dorsiflexors, and lateral trunk flexors; Myofascial release of the quadriceps and dorsiflexors.
Exercises for muscle activation	Movements to activate abdominals and lateral flexors and extensors of the trunk.
Exercises of functionality	Practicing moving around seated on a platform, using the slope of the pool to ease and then hinder, in addition to working at different depths; Working on limits of seated stability with playful activities, being hindered by different support bases and water turbulence.
Cardiorespiratory work	Breathing exercises aimed at greater thoracic mobility; Boosting strength of the respiratory muscles by blowing bubbles using a straw; Low intensity cardiorespiratory conditioning with reciprocating movements of lower limbs while floating.

Table 1. Measure of motor function (MFM)

Dimension	Pre-intervention	Post-intervention
	Score (%)	Score (%)
1 Standing and transfers	2 5.12%	2 5.12%
2 Proximal and axial mobility	30 63.3%	32 66.66%
3 Distal mobility	14 61.9%	15 71.42%
Total score (%) (1+2+3/3)	43.44%	47.73%
Goal (%) (2+3)	62.60%	69.40%

Table 2. Results of the Functional Reach Test, Travel time seated, and the Index of energy expenditure

	Pre-intervention	Post-intervention	Variation in scores
Functional Reach Test (cm) - FRT	6	22	16
Travel time seated (s) - TTS	36	15	-19
Index of energy expenditure (bpm) - IEE	360	107.69	-252.31

cm: centimeter; s: seconds; bpm: beats per minute

and increased activation of the muscles recruited in performing this function.

The measurements used in this case report complemented each other and were responsive to the changes presented by the child during the period. The use of validated and specific tools was of fundamental importance, such as the MFM scale, which enables the evaluation of proximal, distal, and axial motor dysfunctions by means of evidence that is classified in three dimensions, being an important tool for the NMD's showing a degree of sensitivity and good responsiveness, especially in children.¹⁰

A study of patients with Duchenne Muscular Dystrophy (DMD) evaluated changes in the MFM, at an interval of 6 months where there had been no intervention, where dimension D1 showed a lower score because some patients were not ambulatory, and in D2 and D3 there was a slight reduction in the score showing that the scale may be useful for evaluating the progression of the disease, even over a short period of time.¹¹ However, analysis of the results of the D3 for patients who were not ambulatory showed a larger reduction in the score suggesting the progressive involvement of the motor function of the distal limb. These data corroborate the present study because the result that showed the highest percentage of improvement was the D3 (distal mobility), followed by the D2 (proximal and axial mobility). These results are consistent with the focus of the program of aquatic therapy that prioritized the distal mobility of patients with relation to their reaching ability, as well as the proximal and axial mobility in transferring while seated. As expected, the D1 (Standing and transfers) remained with the same score before and after the intervention, because the child did not present motor conditions for this ability.

In typical development, acquisition of upper limb reach is expected to occur around the age of 4 or 5 months. After this acquisition, the processes arise for refining adjustments in proximal reaching, both uni- and bi-manual, as well as in distal, but also the orientation of the hand and opening of the fingers to make contact and seize an object. These skills are impaired in children with CMD, who present proximal weakness that reduces their reaching capacity; this impairment in raising the arms above the shoulder or head interferes with their functional capacity and the implementation of simple activities of daily living such as eating.¹² In the present study the FRT was used to report the reach of the upper limbs, where improvements of 19 cm were observed

in the functional reach. This factor favors the function of picking up objects in front; these gains are attributed to the physical properties of water and to specific maneuvers, because buoyancy favors lifting of the arms and the consecutive recruitment of the shoulder muscles whereby they gain strength.

The TTS and the IEE were calculated to verify the improvement in agility of the patient as well as the amount of energy spent during this movement. Some studies with patients of NMD's report less energy expenditure from children with DMD than from healthy children. Individuals with DMD are known to have decreased muscle activation¹³ and patients with CMD present muscle alterations similar to those with DMD; the results in this case study were contrary to those studies, because the measurement was made during a dynamic activity, and a valid justification of these discordant results is that the stimulated muscle experienced gains in strength and significantly reduced IEE since the start of the program.³

The underwater EMG is one more tool to direct the treatment in this environment. The therapist will have useful feedback for the rehabilitation of the patients, with greater control of exercises performed, and validation of techniques and treatments through quantitative data, which can promote a customized exercise routine.¹⁴ Some studies have used the EMG to analyze abdominal exercises in liquid medium and observed that during the downward phase of the exercise the resistance is multi-directional and the vertical force opposed to gravity calls for an agonist action of the extensor muscles of the vertebral column and thus muscle activity. In this study, due to the conditions of the disease, there was no observable improvement in muscle strength, but the EMG registered greater muscle activation, and this activation is seen as essential for improving the muscle strength of the quadratus lumborum, thereby decreasing the energy expenditure during functional activity.¹³ This is a remarkable, innovative finding, because studies with underwater EMG are rare with NDM patients.

The intervention activities used in this case report and the creation of a specific program based on the concept of the ICF used the physical principles of water, such as movements in the opposite direction of thrust, using the resistance of the water to activate the musculatures recruited in these same functions in a sitting posture. The skills such as seated transfers and reaching for an object in front were initially conducted at a more shallow immersion level and subsequently deepened to

hinder the activities, since the viscosity of water acts as a barrier to the movement, leading to greater resistance. When the subject moves with greater difficulty, there are compensations in muscles and joints, aside from an irregular speed of motion, so the water flow tends to hold the person back, which is why the greater the force to overcome the resistance, the greater the movement and consequent muscle activation.

Several studies show the benefits of aquatic physiotherapy for patients with NMD's, and they are directed not only to quality of life, but to the functionality of the individual. This study observed an improvement in the trunk functionality and functional reach of the child as well as the speed of travel while seated. The premise of the present work is parallel to another study that sought to verify the effects of aquatic therapy on a child with AEP type III;¹⁴ in that study, the main objective was to stimulate functionality and promote the typical development.

In similar studies, some authors have observed an improvement of muscle strength and balance, a reduction of pain, and functional improvements for mobility or motor skills, reporting that these gains in water could be transferred to activities on the ground.¹⁵ The present study observed these transfers through specific tests such as the FRT and the TTS and attributed these gains to maneuvers and exercises that emphasized the activation of musculatures recruited in these functions. Because in the water it is possible to work at different depths, which can be an enabler especially for children who are afraid and lack balance for activities on the ground. The water acts as a protective barrier slowing the response time and enabling the organization of a balance strategy, thus facilitating and increasing their confidence.^{14,15}

Aquatic therapy is an attractive form of therapy and can motivate the child to participate, improve their performance, and increase compliance during a rehabilitation program. The playful aspect should always be emphasized, because water is a stimulating factor for children. This entire therapy program was developed with activities focused on motivation so their wish to participate in the activities encourages the rehabilitation process leading to a more effective therapeutic response.^{14,15}

As limitations of this study, we cannot say that the results were due only to the completion of this aquatic therapy, because the child did other therapies during the period of this protocol. A battery of tests specific to CMD is not available in the literature, nor are

there any similar studies for comparison of results, therefore evaluation instruments are necessary to orient clinical practice and to be a source of guidance for rehabilitation determining the frequency, duration, and intensity of exercises. Future studies should also have larger samples including individuals at other stages of the disease, as well as other study designs such as randomized clinical trials.

CONCLUSION

The results showed that aquatic therapy has been favorable for this patient interfering positively in his functional reach, agility, and index of energy expenditure during travel while seated. The patient presented gains in all the parameters evaluated during the 12-week period showing that, even with the disease's characteristically progressive loss of strength, the child was able to evolve within the functional pattern in which he finds himself.

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