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Physical Therapy Interventions and Response to this Treatment in a 13-year-old Female with Paramyotonia Congenita

[1]Morgan Ricks
[2]Warren Lake
[*3]Trent Jackman

ABSTRACT

Background: Paramyotonia Congenita (PMC) is a rare genetic disorder that affects the sodium ion pump at the level of muscles, retarding muscular relaxation after activation. Symptoms may include isolated or global muscle stiffness, with or without pain. The purpose of this case is to describe physical therapy interventions and response to this treatment in a 13-year-old female with paramyotonia congenita.

Case Summary: 13-year-old female diagnosed with Paramyotonia Congenita and a 4-month history of increased low back and bilateral hip pain. Treatment consisted of therapeutic heating modalities, aquatic therapy, and stretches. Patient and family education and coordination for an individual education plan were also implemented.

Outcome Measures: The patient was treated at an outpatient clinic for 13 sessions. She showed improvements in shoulder, hip, and back range of motion and pain. Goniometric measurements were used to track the range of motion, and a 0-10 visual analog scale and Wong-baker FACES pain rating scale were used to measure pain.

Conclusion: Physical therapy intervention may be an effective treatment option in reducing pain in a 13-year-old girl with PMC. Treatment modalities utilizing heat application may have decreased pain for several days at a time, and deeper heating options appear to yield greater effectiveness at reducing pain for longer periods. However, additional research is needed to evaluate the effectiveness of physical therapy treatment with PMC, including the role and application of deep heat and other treatment modalities.

Keywords: Physical Therapy, Paramyotonia Congenita, Rehabilitation, Intervention, SCN4A gene mutation.

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[1]Doctor of Physical Therapy
Physical therapist, 36 Winn Dr, Rexburg, ID 83440.
email: morganricksdpt@gmail.com
[2]Doctor of Physical Therapy
Director of the NeuroTEAM at Madison Memorial Hospital, 450 E. Main Street, Rexburg, ID 83440.
email: warren.lake@madisonmhealth.org

CORRESPONDING AUTHOR

[*3]Trent Jackman

Doctor of Physical Therapy
Clinical Associate Professor of Physical Therapy,
Idaho State University Campus Box 8045,
Pocatello, ID 83209.
email: trentjackman@isu.edu

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INTRODUCTION

Paramyotonia Congenita (PMC) is a rare genetic disorder that affects the sodium ion pump at the level of muscles, retarding muscular relaxation after activation [1]. The roots of this diagnostic name are derived from myotonia, meaning muscle stiffness and increased resting muscle tone, which ironically occurs during exercise, not after. Therefore, the name 'paradoxical myotonia.' An SCN4A gene mutation causes congenital onset [2] and may lead to specific impairments in the potassium ion pump [1,3].

Symptoms may be detected in the first decade of life and maintained throughout the individual's lifespan. Episodic symptoms in individuals with PMC may last weeks to months [1]. Common symptom triggers include weather or ambient temperature changes, minimal to moderate exercise, or increased functional activity stress [4,5]. McManis et al. demonstrated that they could detect patients with periodic paralysis via electrodiagnostics during exercise tests with repeated contraction [3].

Based on these triggers, individuals with PMC may be at higher risk for sequelae associated with inactivity. Similar concerns may also limit interventions associated with participation in physical therapy interventions or poor outcomes with episodes of physical therapy care.

To date, no peer-reviewed literature outlines physical therapy treatment, or reaction to physical therapy treatment, for individuals with Paramyotonic Congenita. The purpose of this case is to describe physical therapy interventions and response to this treatment in a 13-year-old female with paramyotonia congenita.

Patient Information.

The patient was a 13-year-old female with a positive medical and laboratory diagnosis of Paramyotonia Congenita. The patient's father, one younger sister, and an older brother of the five children family have similar symptoms. The patient reports primary symptoms, associated with this diagnosis, including muscular spasm, acute muscle weakness, and isolated or generalized pain exacerbated by exposure to cold, strength training tasks, and repetitive movement. She states that she has managed these symptoms to avoid exacerbating tasks and environments, with intermittent symptom "flare-ups" several times per year. Resolution to these symptoms within 1-3 days to 2-3 weeks. Decreased activity levels and avoidance of triggering environments alleviate worsening symptoms. Pt with no other significant past medical history.

The patient reports intermittent exacerbation of symptoms associated with school work due to the increased weight of carrying her bookbag between classes.

The patient presented for initial physical therapy evaluation with a 4-month history of increased low back and bilateral hip pain. She had previously been evaluated at a separate facility and participated in a single therapy session (including therapeutic exercise and manual techniques), facilitating significant exacerbation of pain and lethargy. Based on pt's response, she was referred to our physical therapy office for reassessment and treatment.

Examination

Physical therapy examination consisted of range of motion testing, 0-10 subjective report pain scale, assessment of activity limitations, and small-scale demonstration of symptomology using the patient's hand. In addition, separate genetic testing confirming the patient's diagnosis was completed through Primary Children's Hospital in Salt Lake City, Utah, U.S.A.

Subjective reports consisted of a 4-month history of increased low back pain and bilateral hip pain. Symptoms included muscle spasm, acute muscle weakness, generalized pain associated with exposure to cold, repetitive movement, and strengthening tasks. In addition, pain migrated throughout the spine with increased pain at L4-5 and C5-7 bilaterally. The pain was also reported from the superior angle of the scapula to the nuchal lines. Exacerbating conditions include temperature changes, and the patient demonstrated this via holding one hand under cold water for 2 minutes with resultant dystonic movements in hand.

Objective testing revealed a significantly reduced active range of motion bilaterally in the hips and back. Hinging was noted at approximately T9 during trunk flexion. The patient's subjective pain report showed an increased passive range of motion compared to active with an empty end feel. The patient also exhibited a lateral shift during early therapy sessions.

Diagnosis and Assessment

The patient's physical therapy diagnosis was reduced back and hip range of motion secondary to muscle dystonia related to medical diagnoses of Paramyotonia Congenita. Decreased motion and dystonic movements caused pain and reduced function in daily activities. Primary Children's Hospital confirmed the medical diagnosis through Invitae diagnostic testing. A pathogenic variant of SCN4A was noted, associated with various myotonic disorders with hyper or hypokalemic factors, including paramyotonia congenita. A heavy school bag was also associated with increased pain and dystonia. Subjective reports of pain levels were incongruent with objective findings. The patient consistently reported high pain levels in the 8-10/10 range at the beginning of therapy without signs of distress. This finding may have indicated a catastrophizing of pain and possible fear avoidance behaviors.

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Paramyotonia Congenita		
Impairments: Decreased back and hip range of motion Pain	Activity Limitations: Walking Lifting objects Activities of daily living	Participation Restrictions: Difficulty with school and social functions due to pain
Environmental Factors: Individual Education Plan Multiple family members dealing with illness Cold climate	ICD 10 Codes: G71.19 Other specified myotonic disorders	Personal Factors: Young age Catastrophizing symptoms

Interventions

The patient was seen 2x/a week for eight consecutive weeks. Physical therapy interventions consisted of a gentle progressive stretching regimen with pt shoulder to neck deep in a heated therapy pool (99°F, 37.2°C) for 30-45 minutes per session. Stretches were increased with each visit to target back, shoulder, and hip musculature to

promote normalization of muscle tone. Stretching activities in the pool were also done slowly with 3-5 minute breaks between stretches to avoid exacerbation of symptoms. Stretches began with two sets of three repetitions with 3-5 second holds and progressed gradually to two sets of two, with sixty-second holds. Time was decreased between sets of stretches in later visits with no increase in symptomology. The patient had varying responses to intervention but most consistently reported significantly reduced pain or no pain following time in the heated pool environment performing a stretching routine. Therapeutic results from pool activities usually lasted from 1-3 days following therapy and seemed to be influenced by the patient's activities at home and school. Ultrasound set at therapeutic heating levels (1.5 W/cm^2, 3MHz, 5 minutes) was also used to specifically target painful areas beginning with the upper trapezius resulting in reduced pain [6].

Therapy Pool Stretches/Activities	Therapeutic Modalities
Sitting, bilateral knees to chest	Heating provided by Therapy pool
Gentle movement, walking, arm oscillations	Ultrasound
Shoulder flexion and abduction stretches, facilitated by float weights in the pool.	
Across the body shoulder stretches.	
Standing, hip extensor stretches	

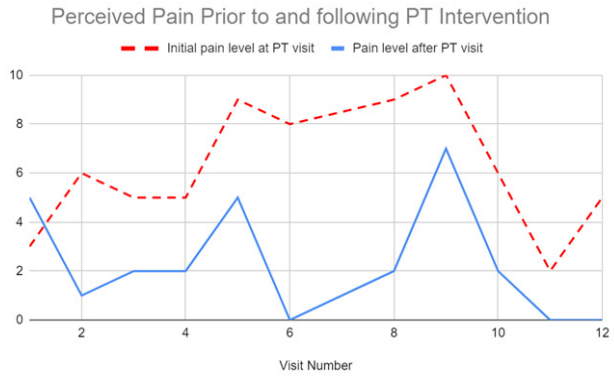
An individual education plan (IEP) was enacted through the school system to allow for adaptations to reduce the number of books and other items carried between classes and to and from school, reducing the weight of the patient's backpack to improve symptoms. The patient was also educated and trained on using bilateral backpack straps instead of the usual unilateral strap support.

Increased compliance with energy conservation techniques and use of the IEP combined with consistent therapy visits and ultrasound as a heating modality allowed longer lasting reductions in the patient's pain. The patient was subsequently discharged from care.

Outcome measures

The patient had a notably limited range of motion, as described in the table below. Motion showed improvement throughout the therapy, but frequent goniometric measurement was discontinued due to the high variance rate between sessions. The patient reported the pain as the limiting factor and had an empty end feel with these measurements. A 0-10 pain scale was consistently used with patient subjective reports ranging from 7-10 at the beginning of therapy to 0-2 following pool activities. A visual comparison of pain values for each visit can be seen in the graph below.

Passive ROM	Evaluation	Third Visit	Fourth Visit	Seventh Visit
Trunk Flexion	29°	52°	75°	20°
Trunk Extension	9°	15°	25°	5°
Shoulder Flexion		L91° R89°	L118° R84°	L82° R73°
Shoulder Abduction		L115° R94°	L136° R126°	L92° R98°
Hip Flexion	L88° R90°	-	L86° R 86°	L96° R105°



DISCUSSION

The patient responded well to physical therapy interventions, with improved pain scores and variable response as determined with ROM. Traditional land-based therapeutic exercise tasks and manual techniques were not attempted with the patient, as previous attempts with these interventions at a separate physical therapy office resulted in the patient's report of increased and prolonged pain, which limited functional performance.

As noted in Table 1, pt demonstrates variable ROM values, including swings of 20-50 deg within sessions. This led the therapist to conclude that goniometry seemed arbitrary and highly swayed by the patient's pain level. Thus the values became a function of pain rather than true joint limitations. Therefore the therapist was unsure of the conclusiveness or consistency of this measure of progression.

The patient was asked to report perceived pain scores on a 0-10 scale at the beginning and conclusion of each physical therapy session. The retrospective analysis notes that days of reported increased pain scores (greater than 8/10) directly corresponded to significant external stressors such as a new trimester in school, disciplinary action taken by parents, or an upcoming holiday with family. In addition, these periods indicate the greatest in-session, perceived gains due to physical therapy interventions. For example, pt reported 8/10 at the beginning of visit 6 and 0/10 pain following 60 minutes of therapy intervention.

The patient's elevated subjective pain reports did not always correspond to observed objective findings. For example, casual conversations with no facial grimace could be performed by pt during reported high pain levels. Therefore, given the patient's juvenile age, the accuracy of her elevated pain reports was questioned. To investigate this question, the Wong-baker FACES pain rating scale was used [7]. This is a 10-point scale with facial icons depicting a simple cartoon face with increasing concern or pain and short matching descriptions. The scale increases from 0/10 (No pain, with smiling figure) to 10/10 (Worst pain possible, crying figure). This scale had been used during previous sessions, but pt chose the face that matched her original subjective number report. To assess the accuracy of the patient's report, the faces from this scale were cut out and placed randomly on a sheet of 8" x 11" 7 paper. Numeric values corresponding to each figure, and its matching description, were removed. Pt was then asked to choose which face most closely matched her current pain

level. For example, the patient had initially reported 9/10 pain at the beginning of therapy. With this modified scale, pt reports 8/10 with the Wong-baker FACES pain rating scale. This one-point difference increased confidence in the patient's subjective reports.

In addition to this subjective measure, the patient demonstrated improved objective findings with physical therapy interventions indicating reduced pain levels. First, the patient's mother reported that since initiating therapy, the patient had not required any outside medication to relieve symptoms or allow for improved movement at home. Previously, the patient had utilized nonsteroidal anti-inflammatory medication several times weekly to control pain. Second, on two separate occasions (visit 2 and 8), the patient presented to physical therapy with a functional 2-3" lateral spinal shift. This shift did not only reduce with cues or distraction only but resolved on both occasions with therapy interventions, as noted above.

The heat was noted to be the most effective treatment to reduce the patient's pain. Initially, the heat was applied with immersion in a heated therapy pool (99°F, 37.2°C), with significant reductions in the patient's reported perceived pain sustained for 1-3 days. Next, a targeted moist heat pack was applied based on pt's good response to warm water. Pt reported enjoying the warmth but denied reduced pain under the selected area. The application of moist heat is found to penetrate at a shallower penetration depth compared to therapeutic ultrasound.[6,8] Therefore, deeper heat via therapeutic ultrasound was then attempted. Unfortunately, pt was poorly positioned (seated with sustained cervical flexion) during the first application of ultrasound, with resultant increased cervical and upper thoracic pain. During the next session, patient positioning was improved with supported head, trunk, and bilateral upper extremities. Due to time restraints, only one side of pt's affected bilateral upper trapezius muscles was treated. This unilateral application allowed for comparison contralaterally. The patient reported a significant reduction in pain, only on the treated side, which was sustained for 13 days. She noted that the treated side felt "lighter." Unfortunately, the patient abruptly discontinued additional therapy intervention shortly after, so this treatment modality's longer-term effects are unknown. Due to the effectiveness of the heating interventions, other pain-reducing treatment modalities, such as electrical stimulation, were not attempted.

The patient was also trained in energy conservation techniques. She reported a directly proportional correlation between her bilateral shoulder and back pain to the size and weight of her school backpack. After coordinating and creating accommodations at the patient's school to reduce the backpack weight and training the patient on using bilateral shoulder straps instead of usual unilateral holds, pt reported significant improvement in pain. These gains were maintained over several weeks with the application of these accommodations.

CONCLUSION

Physical therapy intervention may be an effective treatment option in reducing pain in a 13-year-old girl with PMC. Treatment modalities utilizing heat application may have decreased pain for several days at a time, and deeper heating options appear to yield greater effectiveness at reducing pain for longer periods. However, additional research is needed to evaluate the effectiveness of physical therapy treatment with PMC, including the role and application of deep heat and other treatment modalities. In addition, additional research is needed to assess this population's response to physical therapy treatment and generalized response to these and other treatment interventions.

REFERENCES

[1] Hahn C, Salajegheh MK. Myotonic disorders: A review article. Iran J Neurol 2016: 15(1):46-53.

[2] Huang S, Zhang W[b], Chang X, Guo J. Overlap of periodic paralysis and paramyotonia congenita caused by SCN4A gene mutations two family reports and literature review. Channels 2019; 13(1): 110–119.

[3] McManis, P. G., Lambert, E. H., & Daube, J. R. (1986). The exercise test in periodic paralysis. *Muscle & nerve*, *9*(8), 704–710

[4] Matthews E, Tan SV, Fialho D, Sweeney MG, Sud R, Haworth A, et al. What causes paramyotonia in the United Kingdom? Common and new SCN4A mutations revealed. Neurology 2008; 70(1): 50-3.

[5] Haass A, Ricker K, Rudel R, Lehmann-Horn F, Bohlen R, Dengler R, et al. Clinical study of paramyotonia congenita with and without myotonia in a warm environment. Muscle Nerve 1981; 4(5): 388-95.

[6] Draper DO, Castel JC, Castel D. Rate of Temperature Increase in Human Muscle During 1 MHz and 3 MHz Continuous Ultrasound. J Orthop Sports Phys Ther. 1995;22(4):142-150. doi:10.2519/jospt.1995.22.4.142

[7] Wong DL, Baker CM (1988) Pain in Children: Comparison of Assessment Scales. Pediatric Nursing 14 (1): 9-17).

[8] Ostrowski J, Herb CC, Scifers J, Gonzalez T, Jennings A, Breton D. Comparison of muscle temperature increases produced by moist hot pack and thermostim probe. *Journal of Sport Rehabilitation*. 28(5):459-463. doi:10.1123/jsr.2017-0294