2017

Clinical Implications of Plantarflexion Contracture in a Patient with Epidermolysis Bullosa: A Case Report

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Abstract

Background: Epidermolysis Bullosa is a rare skin disease with the hallmark of extensive blistering and scarring in response to mechanical trauma and friction. Scar formation can result in joint contracture that can affect functional mobility. Movement compensations from the contracture can result in decreased participation and increased pain. Surgical intervention followed by rehabilitation therapy may be required to release the contracture and restore safe functional mobility. ***Case Description:*** A 17-year-old male received physical therapy after surgical release of his right heel cord contracture secondary to a diagnosis of Epidermolysis Bullosa. The patient initially demonstrated a compensatory gait pattern that resulted in decreased endurance and increased knee pain as functional ambulation was regained during the plan of care. ***Intervention:*** Due to the underlying skin disease, the patient could not tolerate typical range of motion restoration techniques including soft tissue mobilization, joint mobilization, or aggressive stretching. Thus, range of motion was addressed through weight-bearing activities. ***Outcome:*** The patient's ankle range of motion improved by 36 degrees, the primary outcome measure assessed. Improvements in functional strength and endurance were observed using the 6-minute walk and 30 second sit to stand tests. ***Discussion:*** Contractures at the ankle are common in patients with Epidermolysis Bullosa due to the nature of the skin disease. Rehabilitation and restoration of motion is complicated by the fragility of the skin. The clinician must be able to implement creative interventions to help restore range and function but also protect the patient from further blistering and scarring. This case presents one approach that was well tolerated and resulted in improved outcomes.

Background

Epidermolysis Bullosa (EB) is a rare genetic connective tissue disorder characterized by blister formation in response to mechanical trauma. Around 200 children are born with EB in the United States per year.[11] There are multiple forms of the genetic disorder, differing according to skin morphology, but all share the same prominent characteristic of fragile skin that blisters with minor friction or trauma. In addition to blistering on outer skin, EB can affect the lining of internal organs and other body systems.[3,11] Those affected by EB are often called "Butterfly Children" because their skin is said to be "as fragile as the wings of a butterfly".[11] The effects of EB are pervasive, debilitating, and painful. Co-morbidities associated with EB include: contractures and scarring that are often treated with surgical release or amputation, anemia from extensive blistering and bleeding, GI and respiratory issues from blistering and chronic inflammation of the structures, cardiomyopathy, genitourinary complications, chronic pain, and cancer.[4,9] As a result, nutrition and development can be affected. There currently is no cure for Epidermolysis Bullosa.

Physical therapists may be able to play an important role in the functional development of children that suffer from EB. Physical therapists can provide patient education for safe setup for developmental mobility from birth through adolescence to minimize shear forces and friction on skin. Patient education and demonstration is often given for compensatory changes for safe mobility including transfers, posture, and mechanics to minimize friction and blister formation. Physical therapists will often encourage and implement exercises and activity in addition to light gentle stretching to prevent or delay almost unavoidable contractures.[9] Hands, feet, and ankles are common areas that are affected by contractures that often require surgical release. Physical therapists then play an important role in maintaining the regained range of motion and promote safe mobility to preserve it.

As Epidermolysis Bullosa is such a rare disorder with complications affecting the musculoskeletal system resulting in impaired function in patients that suffer from it, it is likely that many physical therapists are not aware of indications and contraindications associated with this condition. Thus, the purpose of this case report is to shed light on physical therapy interventions targeting improved function and ROM following surgical heel cord release in patient with this rare skin disorder. Rehabilitation following surgical release in patients that suffer from EB must be approached with great care to ensure good patient outcomes while also protecting the integrity of their skin due to its fragility.

Case Description

The patient is a 17-year-old male high school junior, diagnosed with Epidermolysis Bullosa as an infant. The patient has been a participant in physical therapy throughout his adolescence to promote safe mobility and development through education and range of motion exercise. The patient has also undergone scar tissue releases in both hands for which he participated in occupational therapy to improve function and maintain range of motion. He has regularly participated in novel drug trials for his diagnosis of Epidermolysis Bullosa.

The patient presented to physical therapy after heel cord release on his right Achilles' for a plantarflexion contracture that was affecting his mobility and participation. The plantarflexion contracture began as a blister on the back of his heel. The blister scarred down but was repeatedly re-opened. The cycle of scarring and re-blistering continued over the course of seven years. The process was painful and affected the patient's ability to walk which led to compensatory protective movements that accentuated the contracture such as shortened stride length of the affected leg resulting in decreased ankle dorsiflexion and decreased hip extension on the affected leg resulting in decreased anterior tibial translation. Due to the difficulty with ambulation, the patient opted to complete mobility with a standard manual scooter. He propelled the scooter with his affected leg for household and community mobility. Scooter

propulsion promotes plantarflexion and continued to accentuate the contracture. Just prior to his surgery, the patient's right ankle was contracted to 70° of plantarflexion.

The initial contracture accumulated over the course of 7 years where he was spending less and less time in weight-bearing on his right lower extremity and was ambulating less. Not only did this accentuate the contracture, but the disuse resulted in decrease in functional strength in his right lower extremity, especially in the hip.

At the initial physical therapy evaluation, 2-weeks following his heel cord release, the patient had active dorsiflexion range of motion of 40° short of neutral and passive dorsiflexion range of motion of 29° short of neutral. At initial evaluation, patient was non-ambulatory due to range of motion restriction in the lower extremity that underwent the heel cord release.

Clinical Impression #1

The patient presented to physical therapy with a primary problem of decreased range of motion and functional strength following surgical release of a heel cord contracture. The severe deficits in dorsiflexion range of motion prior to and following surgical intervention hinders the ability of the patient to independently ambulate which affects his participation in school and in the community.

Post-operative management of heel cord release is a typically straightforward rehabilitation process. However, in this case the rehabilitation plan is complicated by the underlying diagnosis of Epidermolysis Bullosa. The patient is a good example of how the rare genetic skin disorder EB can lead to an orthopedic diagnosis and how it can complicate and change an otherwise straightforward rehabilitation plan following a surgical intervention.

Intervention

Restoring and maintaining range of motion is the main purpose of rehabilitation following surgical release of a heel cord contracture. Typical range of motion interventions include soft tissue mobilization, joint mobilization, and stretching to go along with eventual functional strengthening and neuromuscular re-education. This patient's rehabilitation is complicated by the underlying skin disease of Epidermolysis Bullosa. Fragile skin that blisters with friction, the hallmark of the disease, prevents any soft tissue mobilization to be performed. The patient did not tolerate joint mobilization and was hesitant about passive stretching, and only tolerated mild stretching for short periods of time early in the plan of care until he could tolerate weight-bearing on the affected leg.

Progress Examination

As range of motion and functional strength was gained throughout the initial plan of care, more dynamic and functional assessments could be added to later examinations. At a later re-assessment, the patient's passive, active, and weight bearing ankle ranges of motion were assessed with added functional tests including scooter mobility, 6-minute walk test, and 30 second sit to stand.[10] Scooter mobility was an assessment of distance the patient could propel his scooter in a 3-minute time period. The assessment was performed bilaterally and was a measure of endurance and strength of the stance leg. The hope was to promote symmetrical use of each leg during daily scooter propulsion to prevent overuse of a plantarflexion movement on the right leg. The 6-minute walk test was an assessment of gait independence, quality, and endurance. The 30 second sit to stand was used to assess functional lower extremity strength and endurance.

At the progress examination three months into the physical therapy plan of care, the patient had active dorsiflexion range of motion of 18° short of neutral, passive dorsiflexion range of motion of 12° short of neutral, and weight-bearing dorsiflexion range of motion of 5° short of neutral.

The patient demonstrated a Trendelenburg gait with a tendency to hyperextend his right knee during stance phase due to lack of hip abduction strength and decreased ankle range of motion, respectively. He was able to tolerate farther distances with ambulation after regaining some range of motion in his ankle and expressed complaints of mild to moderate persistent right knee pain as he became a more functional ambulator during his recovery.

Clinical Impression #2

As range of motion improves and allows for increased functional mobility and tolerance, gait and functional strength tests will be assessed including: 30 second sit to stand, 6 minute walk test, and a novel assessment of scooter mobility that was specifically relevant to this patient.

At the initial evaluation, the patient would not have been able to participate in the functional assessments. Three months into the rehabilitation process, the patient made good progress with range of motion and was able to be assessed with more functional outcome measures.

As the patient has progressed, he has had the ability to become more active with his participation in activities of daily living and day to day mobility. Though he has become functionally stronger, he still lacked significant dorsiflexion range of motion. He has met the initial goal of household ambulation and is able to negotiate stairs in the home but continues to complete longer distances and most daily mobility with his scooter. As he has progressed independence with mobility, he began to develop pain in his right knee. The lack of dorsiflexion in his right ankle and the lack of strength in his right hip abductors resulted in a Trendelenburg gait with mild hyperextension in right knee. The affect on his gait kinematics and gait pattern from decreased hip strength and poor ankle range of motion has changed biomechanics and forces experienced at the knee.

The progression of the patient led to modified goals to increase ambulation independence and endurance to tolerate community level distances. He added a goal to be able to negotiate stairs with a reciprocal pattern to continue to progress independence with mobility. Management of knee pain and prevention of future knee pain was another added goal. The patient maintained the goal of improving dorsiflexion range of motion in the right ankle to promote increased quality and safety with mobility.

Intervention

Due to the intolerance of friction producing interventions such as soft tissue and joint mobilizations, gains in range of motion needed to be achieved through mild passive range of motion and functional weight-bearing exercises including standing, standing with anterior-posterior weight shifts, step ups with anterior-posterior weight shifts, mini-squats, walking, stair negotiation. The patient participated in physical therapy from two weeks post-op for at least 5 months. He was seen three times per week for four months before dropping down to two times per week. Functional strengthening, functional mobility training, and range of motion exercises including weight bearing activities and mild passive range of motion were cornerstones of treatment after the patient upgraded his goals for improving range of motion, ambulating community level distances, and negotiating stairs with a reciprocal pattern. Strategies for restoration of range of motion that avoided or decreased friction on the skin were implemented. Weight-bearing activities that included mini squats, lunges, step-ups onto box with anterior weight shift were completed to stretch the gastroc-soleus complex and promote anterior translation of the tibia for dorsiflexion range of motion to go along with light manual stretching into dorsiflexion to the patient's tolerance. As his range of motion improved, he was able to progress to higher level functional weight-bearing exercises. He completed a warm up each session including mini squats, anterior-posterior weight shifts in standing, and standing

hamstring stretch. The patient also participated in overground gait and stair training for further weight-bearing with cues for dorsiflexion.

In addition to restorative techniques, the patient received an AFO brace with a heel lift as a compensatory measure to improve gait pattern and address persistent ankle range of motion deficits. Hip strengthening, single limb balance, the use of the heel lift and AFO, in addition to gait training and repetition were tolerated well by the patient to address his goals for a more efficient gait and his goal to decrease his knee pain.

Outcomes

The primary outcome measure assessed in the plan of care was dorsiflexion range of motion of the right ankle. Dorsiflexion range of motion was assessed at initial evaluation, a progress evaluation three to four months into treatment, and at another progress evaluation five to six months into treatment. The patient's dorsiflexion range of motion improved passively from 29° from neutral to 12° from neutral to 4° from neutral at the three assessments respectively. The patient's active dorsiflexion range of motion improved from 40° from neutral to 18° from neutral to 9° from neutral. Dorsiflexion range of motion in weight-bearing was assessed in the two subsequent progress evaluations following the initial evaluation and was measured to be 5° short of neutral both times. An improvement of greater than 6° in ankle range of motion is indicative of meaningful change.[5]

At initial evaluation, the patient had goals of tolerating walking for up to 10-15' as he was unable to tolerate any ambulation or weight-bearing following his procedure. As functional strength and endurance was gained, he met his initial goal and progressed his goal to ambulating community distances. At the first re-assessment he was able to participate in a 6-minute walk test and walked 1020'. In the second re-assessment, he improved his 6-minute walk test distance to 1220'. While cut-off scores or minimally clinically important differences are not established for this patient's age range or diagnosis, an improvement in 200' is significant to the patient as he stated he felt he made improvements in his endurance and walking tolerance at the second re-assessment.

A novel outcome measure of scooter mobility was used to determine improvements in single limb stance and functional strength of his affected leg. While he was progressing with ambulation during the course of treatment, he continued to complete most of his mobility throughout the day with a standard scooter while propelling the scooter with his leg that underwent the surgical procedure. The patient was able to propel his scooter with single limb stance on his affected leg 420' in three minutes at the first progress evaluation and improved to 610' at the second progress evaluation. Comparatively, the patient was able to propel the scooter 965' and 1085' with his unaffected leg in stance at the two progress evaluations.

The final secondary outcome measure was 30 second sit to stand. There is no research for this outcome measure for the patient's age range or diagnosis. The 30 second sit to stand measure is a test for functional strength and endurance of the lower extremities. The patient continued to express improvement in his mobility status as he gained range of motion and strength during treatment session. This was supported by the improvement of his 30 second sit to stand from 12 to 17 between the first and second progress evaluations. This measure of functional strength, in addition to the other measures of functional strength, were not possible at initial evaluation because he could not tolerate weight-bearing on the heel that underwent the heel cord release.

The patient also noted mild to moderate pain in his right knee initially when he progressed to small bouts of ambulation. As his ankle range of motion and leg strength improved throughout treatment, the patient was able to tolerate further distance with ambulation painfree. At the time of his first progress evaluation, the patient had no complaints of pain in his right knee with ambulation or functional mobility.

Discussion

The purpose of this case report was to describe the genetic skin disease Epidermolysis Bullosa, its effect on joint mobility and function, and highlight the implications on the diagnosis on the rehabilitation of an otherwise straightforward plan of care following a surgical procedure. The fragile skin in children that have been diagnosed with EB is susceptible to painful blistering and scarring that leads to contractures. The contractures are accentuated as the patient attempts to protect the blisters to avoid further pain, as was seen in the patient described in this case report. Over the course of seven years, the patient opened and re-opened blisters on his heel that scarred into a plantarflexion contracture at the ankle. As a way to avoid pain and increase function, the patient completed mobility using a scooter that accentuated the contracture until scarring limited his ankle dorsiflexion to 70° short of neutral.

Proper lower extremity kinematics with mobility are dependent upon sufficient range of motion at each joint. Hyper- or hypomobility at a joint will affect movement and forces experienced at the joints surrounding it that may affect function and lead to injury. A joint contracture is the inability of a joint to move through its full range of motion and its excessive resistance to passive range of motion. Increased stiffness and decreased extensibility of tissues surrounding the joint lead to a joint contracture. Plantarflexion contractures at the ankle joint can affect knee biomechanics, gait mechanics, and lower extremity injury incidence in young children.[1,2,6]

For proper anterior tibial translation during stance phase of gait, the ankle must be able to achieve 10* of dorsiflexion.[6] Decreased motion at ankle joint will often lead to increased motion at joints around it. Most typically, lack of ankle mobility during gait will result in either knee hyperextension or excessive knee flexion through stance phase. Increased knee flexion angle and increased knee flexion moments occur with a knee flexion compensation. When ankle contracture is compensated with knee extension, decreased extension moment and decreased knee power generation occurs.[1,2] Decreased hip extension may also result during stance phase. The effects of contracture on stance phase may affect gait pattern on the contralateral leg resulting in premature initial contact and decreased step length. During swing phase, gait deviations may occur at the hip joint including hitching of the pelvis and increased hip abduction.[1] Increased torque on joints lead them vulnerable to overuse injuries especially during repetitive activities such as gait. Inconsistent and asymmetrical gait patterns are inefficient that lead to increased energy expenditure.

In a 2016 study from Liu and Xie, correlation of lower extremity injury and Achilles' tightness in children was researched.[7] The cross-sectional study could not determine causation of lower extremity injury and Achilles' tightness but was able to demonstrate a significant correlation between decreased dorsiflexion range of motion and traumatic lower extremity injury. The patient did not report any falls or trauma during the course of treatment but this finding is significant for the population of patients that suffer from Epidermolysis Bullosa due to their fragile skin and their increased susceptibility to painful injury in the event of even minor trauma.

Plantarflexion contracture can lead to a vicious cycle of injury, pain, and decreased mobility that leads to further contracture for children that suffer from Epidermolysis Bullosa. Skin blistering and scarring down is common with this skin disease that often leads to contractures of joints. Plantarflexion contracture of the ankle joint can lead to changes in knee and hip kinematics, placing more strain on those joints leading to pain. The changed gait kinematics are often inefficient and energy costly. There also may be a correlation between lower extremity traumatic injury and decreased range of motion in dorsiflexion. Children with Epidermolysis Bullosa are already predisposed to injury due to the nature of the disease. Pain, increased energy expenditure, and increased incidence of injury can lead to decreased participation and fear of movement which may lead to further contracture that requires a surgical release.

Decreased mobility at the ankle joint affects the other joints in the lower extremity. Gait kinematics are changed that result in changes experienced at the hip and knee and lead to an inefficient gait and an increase in forces experienced.[1,2,6] Inefficiencies in the gait will affect endurance and the change in joint forces can result in pain.[1] In this case, the patient compensated for his decreased ankle mobility with mild knee hyperextension, decreased hip extension, and hip abduction with pelvic drop. These compensations likely lead to his complaints of right knee pain as he began to tolerate more walking. He demonstrated decreased endurance with ambulation as well as he could not tolerate increased distances during gait training without taking a seated rest break.

In addition to range of motion, the patient also had goals to decrease pain in his knee, improve his gait pattern and endurance to allow for community ambulation and participation. General strengthening and power exercises in conjunction with functional strengthening that served the purpose of weight-bearing range of motion activities were included in the plan of care. As lower extremity strength was gained and his gait pattern improved, the knee pain resolved. He was able to improve his endurance as evidenced by the 6-minute walk test and subjective reports of community ambulation.

Physical therapy care for the rare skin condition, Epidermolysis Bullosa, is uncommon in the literature. The complications in the musculoskeletal system that lead to decreased functional mobility and participation are well suited for physical therapy interventions to encourage healthy and safe mobility. However, the therapist has the unique challenge of implementing effective interventions to restore functional range and strength without causing blistering and further scarring. This case report describes several strategies used to treat a patient with EB following heel cord release with stretching and strengthening exercises safely without leading to skin breakdown. Further research in this patient population would be useful in the area of preventative strategies to avoid blistering and promote safe mobility.

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