Aquatic Therapy for a Child with Nail Patella Syndrome

A Capstone Project for PTY 768
Presented to the Faculty of The Sage Colleges
School of Nursing and Health Sciences

In Partial Fulfillment
of the Requirements for the Degree of
Doctor of Physical Therapy

Janelle L. Cassant, SPT
May, 2007

Approved:

Laura Z. Gras, PT, DSc, GCS
Research Advisor, PTY 768

Marjane Selleck PT, MS, PCS
Program Director, Doctor of Physical Therapy Program
Aquatic Therapy for a Child with Nail Patella Syndrome
Janelle Cassant
The Sage Colleges
Abstract:
**Introduction:** Nail Patella Syndrome is a rare genetic disorder that is characterized by nail, skeletal, ocular, and renal abnormalities. The severity of the disorder varies between patients. Patients with skeletal abnormalities may need physical therapy treatment to help with functional mobility. Aquatic therapy has been found to help improve functional mobility while decreasing stress on specific joints. **Case description:** Justin is a 2 year old male with Nail Patella Syndrome receiving physical therapy on land and aquatic therapy. **Examination:** Standardized test scores using the *Bayley Scales of Infant Development – 2nd edition* determined significant gross motor delay. The patient scored below the 1st percentile of children his age. The patient’s primary means of transportation was creeping. **Evaluation:** Patient demonstrated gross motor delays; he was unable to pull to stand, stand or ambulate independently. **Interventions:** Aquatic therapy program focused on increasing range of motion, strength and balance while improving gait. **Outcomes:** The patient demonstrated significant improvement in gross motor skills over an 18 month period. **Discussion:** An aquatic therapy program may be beneficial for a patient with Nail Patella Syndrome to improve functional mobility.
Introduction

Nail Patella Syndrome (NPS), also referred to as Turner Syndrome, arthrodysplasia, Touraine Syndrome, hereditary osteoonychodysplasias (HOOD), Fong’s Disease, or Turner-Kieser Syndrome, a rare autosomal dominant genetic disorder that affects tissues of both the mesoderm and ectoderm. NPS is caused by a mutation in the gene LMX1B, this gene is responsible for regulating type IV collagen expression. The gene is considered pleomorphic because it is capable of generating a variety of anomalies.¹ The syndrome is characterized by symmetrical nail, skeletal, ocular, and renal abnormalities.² The incidence of NPS is approximately 1 in 50,000 live births affecting females 10% more often than males.² Severities of clinical features is variable within and between families.² The syndrome demonstrates complete penetrance, being that each individual that carries the gene is affected to some degree.¹ The first attention was focused on this disorder by Little in 1897, who described 42 cases with congenital absence of the patella, or underdeveloped patella.¹ Nail and skeletal deformities are most common in patients with NPS, these features occur in 90-95% of patients.¹ Patellar aplasia or hypoplasia occurs in 92.7% of patients with NPS.³ A patient with a small or dysplastic patella may cause the lateral femoral condyle to wear away causing chronic subluxation or dislocation of the patella.³ Weight-bearing can cause excessive stress on the joints making it difficult for patients to stand and ambulate. An aquatic therapy program may be beneficial for these patients to decrease stress placed on the joints.

Aquatic therapy provides a full body workout without putting excessive stress and tension on specific unstable joints that experience abnormal loading.⁸⁻⁹ Water buoyancy decreases gravitational influences and provides postural support to allow patients to
initiate independent movement patterns they are unable to perform on land.\textsuperscript{8,9} Aquatic therapy programs are common for children with neuromotor impairments. The goal of an aquatic therapy program is to improve activities of daily living and improve body functions.\textsuperscript{8}

The majority of research on aquatic intervention targets the adult population.\textsuperscript{8} Aquatic therapy research for the pediatric population focuses on children with cerebral palsy and other neuromotor impairments. Studies on children with cerebral palsy found significant improvement in flexibility, respiratory function, muscle strength, gait and gross motor function.\textsuperscript{9} A review of 11 research articles on aquatic interventions for children with neuromotor impairment found improvements in body functions, gross motor and fine motor skills, gait and coordination; two of these articles reviewed found improvements in communication skills, social interaction and self-esteem.\textsuperscript{8}

After a review of the literature, it is hypothesized that a patient with NPS may benefit from an aquatic therapy program to improve activities of daily living and gross motor skills. The purpose of this case report is to provide a description of a patient with NPS and describe an aquatic therapy program that was performed to help increase the patient’s functional mobility.

Case Description

\textit{History}

Justin is a 2-year-old male diagnosed with NPS at birth. His mother reported no use of tobacco products or alcohol consumption during the pregnancy. Justin was born vaginally at 41 weeks gestation. He weighed 8 pounds and 10 ounces at birth. Justin was born with absence of nails on his thumbs, bilateral clubfeet with the left leg externally rotated at knee and ankle. In September 2003 he underwent an Achilles tendon release.
He was referred for physical therapy services when he was 2 years old secondary to gross motor delay. Justin was unable to stand or ambulate at this time. He had not received any physical therapy prior to this referral. He underwent 2 orthopedic surgeries during his first year of receiving physical therapy. The first in October 2005 was a re-alignment of his right lower extremity and the second surgery was performed in April 2006 for a re-alignment of his left lower extremity. At this time he is being monitored regularly for glaucoma, however this is not a current concern. Justin’s family has a history of NPS, clubfoot, learning disabilities, and speech delays.

Justin lives with his parents. His parents are both employed outside of their home. Physical therapy is provided through early intervention services. Justin’s support consists of his parents and his grandparents who occasionally care for him during the day when his parents are working.

Justin was chosen for this case report because NPS is a rare genetic disorder. There is not a lot of literature on this diagnosis and almost no information on physical therapy interventions. The Sage College’s Institutional Review Board approved this case report.

*Examination*

On July 27, 2005 a pediatric psychologist and a physical therapist evaluated Justin. He did not demonstrate cognitive concerns; he interacted well with examiners and demonstrated curiosity and interest in tasks that were asked of him. Justin’s cognitive abilities were assessed using the *Bayley Scales of Infant Development – 2nd edition* (BSID-II). The test evaluated the skills such as memory, problem solving, and early concept development. Justin received a Mental Development Index of 104; which falls
within the average range (average range = 85-114) of cognitive development. He was able to follow directions, complete puzzles, and match and name several colors. The BSID-II test manual contains information on reliability of the Mental, Motor, and Behavioral Rating Scales. The BSID-II has high reliability r-values ranging from 0.88 to 0.84.\textsuperscript{11}

Justin’s social-emotional development was assessed using clinical observations, parent report and the \textit{Developmental Assessment of Young Children (DAY-C) Social-Emotional Subtest}.\textsuperscript{12} The DAY-C has a high reliability with coefficients ranging from 0.90 to 0.99 and a standard error of measurement ranging from 1.5 to 4.74.\textsuperscript{12} He received a standard score of 100, which falls within the average range for the patient’s age (average ranges from 90-100). He demonstrated age-appropriate play behaviors and was able to separate from his mother.

Justin’s speech and language was evaluated using the clinical observations, an item analysis of the BSID-II and parent report. He was verbal and used a variety of words and phrases. Justin used an open-mouthed posture and substituted or dropped the initial sound making some words difficult to understand. His speech and language fell within the average ranges.

Justin was initially examined using the standardized BSID-II. This assessment compared his gross motor skills to typically developing children of the same age. It looked at both gross motor and fine motor development. Justin received a psychomotor developmental index (PDI) score of less than 70 (average range is 85-114). Justin’s PDI demonstrated a significant delay in gross motor skills. His performance was 2 standard
deviations below the mean. Professional judgment and standardized test scores indicate
gross motor delay. Refer to Table 1 for the standardized examinations’ findings.

Table 1: Test Findings from Initial Evaluation Using Justin’s Chronological Age of 24
Months.

<table>
<thead>
<tr>
<th>Test</th>
<th>Subtest</th>
<th>Score</th>
<th>Average Score*</th>
</tr>
</thead>
<tbody>
<tr>
<td>BSID-II</td>
<td>Mental Development</td>
<td>104</td>
<td>85-114</td>
</tr>
<tr>
<td>DAY-C</td>
<td>Social-Emotional</td>
<td>100</td>
<td>90-100</td>
</tr>
<tr>
<td>BSID-II</td>
<td>Psychomotor Development</td>
<td>&lt;70</td>
<td>85-114</td>
</tr>
</tbody>
</table>

*Average score for children the same age at the patient

A core evaluation was conduction on Justin. A core evaluation is a
comprehensive evaluation of the patient to evaluate each of the following domains:
physical development, cognitive development, communication development, social or
emotional development and adaptive development. During the core evaluation Justin sat
on the floor with his left foot under his bottom and his right lower extremity positioned to
the right of his body with his knee and ankle bent in a modified W-sit. His medial right
thigh, lower leg, and foot rested on the floor. Justin was able to maintain this sitting
position without using his upper extremities for support or balance. From sitting Justin
was able to transition easily onto his hands and knees to crawl. He was able to crawl
quickly to negotiate his home. Justin crawled with his right hand fisted and weight
bearing on the knuckles of his right hand. This position is thought to be a compensatory
movement pattern to clear his right lower extremity when bringing it forward to crawl.
Justin also demonstrated the ability to walk on his knees. He was able to pull to stand at
furniture by lifting himself onto his left lower extremity and lower himself down with good control onto his right knee using a wide half-kneel pattern. When weight-bearing Justin maintained bilaterally flexed knees (right knee flexion greater than left knee flexion) and externally rotated his right lower extremity secondary to the abnormal alignment of the bones in his lower extremities associated with NPS. Justin’s mother reported that he is able to cruise along the couch in either direction. Justin’s ability to cruise was not observed. Justin attempted to rise to standing from a quadruped position by transitioning onto his feet, weight shifting backwards over his lower extremities and then rising upright. He was able to transition into a semi-standing position briefly with his knee bent until his right knee buckled forcing him to fall to the floor onto his knees. Walking without assistance was not assessed to avoid placing stress on Justin’s knees.

Justin’s fine motor skills were assessed during the core evaluation. He used his eyes and bilateral hands to explore his environment and perform tasks with various items that were given to him. He was able to pick up and hold items his palm and out away from his palm with his thumb and fingers depending on the size of the item. Justin was able picked up 10 pellets using a pincer grip and place them into a container within 60 seconds. He was able to pick up a pencil with the pads of his right thumb and finger. He scribbled with the pencil in his right hand and was able to hold the paper in place with his left hand. Justin demonstrated the ability to stack blocks with accuracy and controlled release of one block on top of the other.

**Evaluation**

Justin demonstrated gross motor delays. He was unable to stand or ambulate independently. Justin demonstrated decreased tolerance to lower extremity weight
bearing. He was unable to pull to stand from sitting. He demonstrated decreased upper and lower extremity strength during transitional movements. His fine motor skills were age appropriate.

Diagnosis

Justin presents with impaired motor function and sensory integrity. The practice pattern appropriate for this patient is Neuromuscular Practice Pattern 5B: Impaired Neuromotor Development. The most appropriate ICD-9 code is 754: congenital musculoskeletal deformities.¹⁸

Prognosis

Justin is a good candidate for physical therapy because he is young and motivated with good family support. It is predicted that he will demonstrate the optimal highest level of functioning at home and in school with gradual improvements in strength, balance, and functional mobility.

Plan of Care

Interventions for physical therapy include patient/family education, home exercise plan, stretching, strengthening, balance training, gait training, and aquatic therapy. Justin was seen 2-3 times a week for 30 minute sessions until he demonstrated safe and independent functional mobility, which would allow him to negotiate in his school and community environment.

Physical Therapy Goals:

Annual Goal: Justin will be able to negotiate within his school, home and community independently, safely, and efficiently.
Objectives:

1. Justin will be able to walk independently 50 feet with good balance and control on even and uneven surfaces while using an age appropriate gait pattern.

2. Justin will be able to run 20 feet on even and uneven surfaces independently with good balance and control.

3. Justin will be able to ascend and descend a flight of stairs with the aid of a rail using an alternating pattern independently with good balance and control.

4. Justin will be able to transition from a sitting position on the floor to a standing position through an age appropriate 1/2 kneel position independently.

Annual Goal: Justin will be able to participate in age appropriate games with his peers.

1. Justin will be able to jump forward with 2 feet 12 inches independently with good balance.

2. Justin will be able to stand on one foot (right and left) for 3 seconds with abducted upper extremities without losing his balance.

3. Justin will be able to kick a playground size ball 10 feet independently while maintaining his balance with less than 45 degrees deviation 75% of the time.

4. Justin will be able to stop rolling a playground size ball with either his right or left foot without losing his balance 3 out of 4 trials.

Interventions

Justin’s aquatic therapy program was followed for 12 months with a re-evaluation after 6 months of intervention and at 18 months after interventions began. The aquatic interventions were temporary on hold twice throughout this case report. In October 2005,
Justin had an orthopedic re-alignment of his right lower extremity; he was unable to participate in an aquatic program for a month while he had a cast on. In April 2006, Justin had an orthopedic re-alignment of his left lower extremity and was unable to participate in aquatics for another month. During both time periods, Justin continued to receive physical therapy on land.

Initiation of an aquatic program began with relaxation techniques in the pool. The physical therapist would begin treatment by applying rhythmical movement at the obliques and trunk to relax the pelvic and hips. The physical therapist worked on weight bearing by giving moderate assistance at the gluteals and obliques to assist standing and lateral weight shifting. Justin performed toe touch weight bearing to improve lower extremity tolerance to weight bearing. Justin then performed a prone swim with moderate assistance at the obliques and gluteals and moderate verbal cueing to promote alternating kicking to increase strength and range of motion. These interventions were performed once a week in the pool for 6 months. Assistance level gradually decreased as the patient’s strength and range of motion improved.

After 6 months Justin became more tolerable of weight bearing with minimal assistance was needed. A walking program was added to his aquatic therapy program. Justin needed moderate assistance at his gluteals and quadriceps to promote lateral weight shifting. To increase Justin’s upper extremity strength pull-ups at the end of the pool were added to the program. Justin needed minimal assistance at his trunk to perform pull-ups. As his upper extremity strength increased, less assistance was given. The walking program and pull-ups were added to the exercises Justin was already performing. Justin had surgery in April 2006 and aquatic therapy was temporarily discontinued.
In May 2006, Justin was able to begin aquatic therapy again. The aquatic program focused on walking and weight shifting over his right lower extremity. Stair training was added to the aquatic program. Justin received moderate assistance at his quadriceps and gluteals as he worked on stepping up and step from a 3-inch step in the water. Assistance decreased as Justin was able to tolerate increased weight bearing on right lower extremity. Justin continued to work on prone swimming without assistance and minimal verbal cues to promote alternate kicking and performed wall pull-ups independently.

In July 2006, jumping and unilateral stance activities were added to Justin’s program. Justin performed two foot jumping in water with assistance from the physical therapist at his trunk to promote abdominal control. Justin was able to walk independently in the pool. The aquatic program began to focus on unilateral stance activities. Justin was able to stand on one lower extremity while shooting a basketball.

All aquatic interventions were carried over onto land. Justin received physical therapy interventions for a total of three times a week. The interventions described above would vary each session secondary to Justin’s attitude, energy level, and tolerance to activity on the day of treatment. Refer to Table 2 for a description of the aquatic therapy program performed with Justin.
Table 2: Aquatic Therapy Interventions with Progression

<table>
<thead>
<tr>
<th>Date</th>
<th>Frequency</th>
<th>Intervention</th>
<th>Purpose</th>
</tr>
</thead>
<tbody>
<tr>
<td>August 2005</td>
<td>1x per week</td>
<td>Relaxation techniques, toe touch weight bearing, prone swim</td>
<td>Increase weight acceptance, increase strength, increase range of motion</td>
</tr>
<tr>
<td>January 2006</td>
<td>1 x per week</td>
<td>Walking program, upper extremity pull-ups, prone swim</td>
<td>Improve weight bearing and ambulation, Increase upper extremity strength</td>
</tr>
<tr>
<td>May 2006</td>
<td>2x per week</td>
<td>Walking, prone swim, pull-ups, step-ups</td>
<td>Improve balance, gait and stair training</td>
</tr>
<tr>
<td>July 2006</td>
<td>2x per week</td>
<td>Step-ups, two foot jumping, unilateral stance</td>
<td>Improve balance</td>
</tr>
</tbody>
</table>

Outcomes

Justin’s gross motor skills were reassessed 6 months after his core evaluation. Justin’s primary means of mobility after 6 months of physical therapy intervention was creeping on his hands and knees with his brace on his right lower extremity. When creeping, Justin demonstrated internal rotation of his bilateral shoulders (right greater than left), right foot pronation and eversion, and left hip internal rotation. Justin was able to transition from quadruped to a sitting position by weight shifting over his left hip onto his bottom. He sat on the floor with his right lower extremity extended out in front of him with mild hip internal rotation and his left lower extremity in a “W” position. Justin was able to transition into quadruped from sitting by rolling onto his stomach, pushing up onto his hands and knees, and shifting his weight over his left hip to a quadruped position. He was able to maintain a tall kneeling position with good balance for about 4-5 minutes.
Justin was able to pull himself to stand using a support surface by positioning his right lower extremity forward with his hip externally rotated and quickly shifted his weight over his right lower extremity to unweight his left lower extremity to move it underneath him. He primarily used his upper extremities to pull to stand. Standing at a support surface, Justin presented with right lower extremity external rotation, right foot pronation and eversion, left hip internal rotation, knee flexion, and plantar flexion. He was able to stand at a support surface without leaning his body on the surface for support.

Justin was able to ascend a flight of stairs by stepping with his right foot first onto a step and shifting his weight briefly over his right lower extremity and upper extremity to step onto his left lower extremity. Justin descended a flight of stairs by lowering his right lower extremity onto a step below him and weight shifting over his right lower extremity briefly to step with his left lower extremity.

Justin was able to throw a tennis ball overhand with good shoulder rotation and direction 7-10 feet through the air. He was able to throw a tennis ball underhand with shoulder internal rotation and abduction with fair direction 5-7 feet through the air. Justin was able to catch a playground size ball with his arms by bending his elbows and trapping the ball with his chest.

Justin presented with normal forward, backward, and lateral protective reactions. He present with an incomplete, spontaneous and sustained forward righting reaction. Justin could maintain his head at midline. When pulling to a sitting position from supine, Justin presented with an initial head lag for the first ½ of the movement. He was able to tuck his chin at the last ¼ of the movement.
Justin’s gross motor skills were reassessed 6 months after his initial evaluation using the Peabody Developmental Motor Scales-2nd edition. The PDMS-2 Gross Motor Section measures gross motor skills of children from birth to 72 months. Refer to Table 3 for his scores on the Peabody at 30 months.

Table 3: Justin’s Score on the Gross Motor Section of PDMS-2 using his Chronological age of 30 Months.

<table>
<thead>
<tr>
<th>Subsets</th>
<th>Age Equivalent</th>
<th>Percentile</th>
<th>Standard Scores</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reflexes</td>
<td>10 Months</td>
<td>*</td>
<td>*</td>
</tr>
<tr>
<td>Stationary</td>
<td>8 Months</td>
<td>25%</td>
<td>8</td>
</tr>
<tr>
<td>Locomotion</td>
<td>9 Months</td>
<td>&lt;1%</td>
<td>1</td>
</tr>
<tr>
<td>Object Manipulation</td>
<td>12 Months</td>
<td>1%</td>
<td>3</td>
</tr>
<tr>
<td>Average Score</td>
<td>13 Months</td>
<td>&lt;1%</td>
<td></td>
</tr>
</tbody>
</table>

* Denotes inability to score secondary to age
Gross Motor Quotient = 61

The sum of Justin’s gross motor scores places him below the 1st percentile of children in his age group. The content of each subtest is as follows: Reflexes-“Measures the child’s ability to automatically react to the environmental events”, Stationary-“Measures the child’s ability to sustain control of his or her body within its center of gravity and retain equilibrium”, Locomotion-“Measures the child’s ability to transport his or her body from one base to another”, Object Manipulation-“Measures the child’s ability to throw, catch, and kick balls”. 11
Concurrent validity between the BSID-II and the PDMS-2 are high. The correlation between the fine motor measurements is 0.87 and 0.83 for the gross motor measurements. Over the first 6 months of receiving physical therapy services Justin demonstrated an increase in protective reactions, righting reactions, and upper extremity strength as evidence by increase in ability to transition in and out of positions. He continues to demonstrate gross motor delays and decrease in strength and muscle endurance in his abdominal muscles and lower extremities.

Justin’s most recent evaluation was conducted in January 2007 using the PDMS-II. He demonstrates improvements in all areas of development. Currently Justin’s primary means of mobility is ambulating independently with moderate gait deviations and decreased balance. Justin walks with a heel to toe gait pattern with a slightly increased base of support secondary to decreased ability to weight shift laterally. He is able to transition from standing to squatting independently with a wide base of support and right lateral weight shift. He is able to maintain a squatting position for a brief period of time before falling to his right side or backwards onto his bottom.

Justin is able to ascend and descend one flight of stairs using a marking pattern with the support of one rail. He is attempting to jump with 2 feet at this time. He is able to jump by leading with his left foot with fair balance. Justin is able to throw a tennis ball overhand and underhand with fair direction. He is able to kick a soccer ball with fair balance a distance of 5-10 feet. He has demonstrated an increase in self-confidence, protective reactions, righting reactions, upper extremity strength, lower extremity strength, balance, and muscle control. Justin continues to demonstrate significant delay
in locomotor skills and a significant decrease in abdominal and lower extremity strength and muscle endurance. Refer to Table 4 for Justin’s scores on the Peabody at 42 months.

<table>
<thead>
<tr>
<th>Subsets</th>
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<th>Percentile</th>
<th>Standard Scores</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reflexes</td>
<td>*</td>
<td>*</td>
<td>*</td>
</tr>
<tr>
<td>Stationary</td>
<td>35 Months</td>
<td>16%</td>
<td>7</td>
</tr>
<tr>
<td>Locomotion</td>
<td>22 Months</td>
<td>2%</td>
<td>4</td>
</tr>
<tr>
<td>Object Manipulation</td>
<td>36 Months</td>
<td>25%</td>
<td>8</td>
</tr>
<tr>
<td>Average Score</td>
<td>31 Months</td>
<td>5%</td>
<td></td>
</tr>
</tbody>
</table>

* Denotes inability to score secondary to age
Gross Motor Quotient = 76

At 30 months of age Justin scored below the 1st percentile of children in his age group, at 42 months of age Justin scored below the 5th percentile of children in his age group.

At the time of Justin’s evaluation in January 2007 he had not met any of his physical therapy goals, however he is not able to walk independently with fair balance, he can negotiate stairs with a step to pattern, and he is able to maintain unilateral stance for 2-3 seconds with fair balance. Justin is currently continuing physical therapy to meet his goals.

**Discussion**

NPS has not been examined well in the literature. This case report gives information on the disease and aquatic interventions that can be performed with this
population of patients. The patient demonstrated improvement with activities of daily living and gross motor skills. Previous research has suggested that an aquatic therapy program can help improve strength, balance, coordination, gait and activities of daily living. The patient in this case report underwent 2 surgeries while receiving physical therapy and he was receiving physical therapy on land as well. Therefore it is difficult to state that aquatic therapy alone would be beneficial for this patient. In the future more research could be targeted toward aquatic therapy alone. During the course of therapy for this patient, the interventions varied from session to session depending on the patient’s attitude that day. More consistent sessions would allow researchers to evaluate which aquatic exercises may be more beneficial to the patient. Evaluation for this case report consisted of standardized test scores and professional judgment. Strength and range of motion testing may have been appropriate to compare results over time.

Overall the patient showed improvement in all areas of development. He is currently receiving physical therapy. The patient received 2 aquatic sessions a week and one land session. He continues to demonstrate gross motor delays when compared to children of the same age, however when compared to himself he has demonstrated significant improvement.

It is suggested that future research should focus on aquatic therapy without a land program, compared to a land program alone. This would investigate the benefits of an aquatic program in comparison to a land program to see which may provide a better intervention for these patients. It would be beneficial to look at more long-term effects of therapy for patients with NPS.
References:

March 13, 2007

Janelle L. Cassant
113 Colleen Road Apt 7
Troy, NY 12180

IRB PROPOSAL # 080-06
Reviewer: Dr. Samuel W. Hill, PhD

Dear Ms. Cassant:

I have completed the review of your project entitled “Balance Training and pool therapy for a child with Nail Patella Syndrome” and your project had been approved. Please disregard any prior correspondence regarding a withdrawal. This was sent in error. Good luck with your research.

Please let me know if you have any questions. Please refer to your IRB Proposal number whenever corresponding with us whether by mail or in person.

Sincerely,

[Signature]

Samuel W. Hill, PhD
Chair, IRB

SWH/rd

Cc: Dr. Laura Z. Gras
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SAGE GRADUATE SCHOOL

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Student(s) Signature / Date
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Introduction

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initiate independent movement patterns they are unable to perform on land.\textsuperscript{8-9} Aquatic therapy programs are common for children with neuromotor impairments. The goal of an aquatic therapy program is to improve activities of daily living and improve body functions.\textsuperscript{8}

The majority of research on aquatic intervention targets the adult population.\textsuperscript{8} Aquatic therapy research for the pediatric population focuses on children with cerebral palsy and other neuromotor impairments. Studies on children with cerebral palsy found significant improvement in flexibility, respiratory function, muscle strength, gait and gross motor function.\textsuperscript{9} A review of 11 research articles on aquatic interventions for children with neuromotor impairment found improvements in body functions, gross motor and fine motor skills, gait and coordination; two of these articles reviewed found improvements in communication skills, social interaction and self-esteem.\textsuperscript{8}

After a review of the literature, it is hypothesized that a patient with NPS may benefit from an aquatic therapy program to improve activities of daily living and gross motor skills. The purpose of this case report is to provide a description of a patient with NPS and describe an aquatic therapy program that was performed to help increase the patient's functional mobility.

\textbf{Case Description}

\textit{History}

Justin is a 2-year-old male diagnosed with NPS at birth. His mother reported no use of tobacco products or alcohol consumption during the pregnancy. Justin was born vaginally at 41 weeks gestation. He weighed 8 pounds and 10 ounces at birth. Justin was born with absence of nails on his thumbs, bilateral clubfeet with the left leg externally rotated at knee and ankle. In September 2003 he underwent an Achilles tendon release.
He was referred for physical therapy services when he was 2 years old secondary to gross motor delay. Justin was unable to stand or ambulate at this time. He had not received any physical therapy prior to this referral. He underwent 2 orthopedic surgeries during his first year of receiving physical therapy. The first in October 2005 was a re-alignment of his right lower extremity and the second surgery was performed in April 2006 for a re-alignment of his left lower extremity. At this time he is being monitored regularly for glaucoma, however this is not a current concern. Justin’s family has a history of NPS, clubfoot, learning disabilities, and speech delays.

Justin lives with his parents. His parents are both employed outside of their home. Physical therapy is provided through early intervention services. Justin’s support consists of his parents and his grandparents who occasionally care for him during the day when his parents are working.

Justin was chosen for this case report because NPS is a rare genetic disorder. There is not a lot of literature on this diagnosis and almost no information on physical therapy interventions. The Sage College’s Institutional Review Board approved this case report.

Examination

On July 27, 2005 a pediatric psychologist and a physical therapist evaluated Justin. He did not demonstrate cognitive concerns; he interacted well with examiners and demonstrated curiosity and interest in tasks that were asked of him. Justin’s cognitive abilities were assessed using the Bayley Scales of Infant Development – 2nd edition (BSID-II). The test evaluated the skills such as memory, problem solving, and early concept development. Justin received a Mental Development Index of 104; which falls
within the average range (average range = 85-114) of cognitive development. He was able to follow directions, complete puzzles, and match and name several colors. The BSID-II test manual contains information on reliability of the Mental, Motor, and Behavioral Rating Scales. The BSID-II has high reliability r-values ranging from 0.88 to 0.84.\textsuperscript{11}

Justin’s social-emotional development was assessed using clinical observations, parent report and the Developmental Assessment of Young Children (DAY-C) Social-Emotional Subtest.\textsuperscript{12} The DAY-C has a high reliability with coefficients ranging from 0.90 to 0.99 and a standard error of measurement ranging from 1.5 to 4.74.\textsuperscript{12} He received a standard score of 100, which falls within the average range for the patient’s age (average ranges from 90-100). He demonstrated age-appropriate play behaviors and was able to separate from his mother.

Justin’s speech and language was evaluated using the clinical observations, an item analysis of the BSID-II and parent report. He was verbal and used a variety of words and phrases. Justin used an open-mouthed posture and substituted or dropped the initial sound making some words difficult to understand. His speech and language fell within the average ranges.

Justin was initially examined using the standardized BSID-II. This assessment compared his gross motor skills to typically developing children of the same age. It looked at both gross motor and fine motor development. Justin received a psychomotor developmental index (PDI) score of less than 70 (average range is 85-114). Justin’s PDI demonstrated a significant delay in gross motor skills. His performance was 2 standard
deviations below the mean. Professional judgment and standardized test scores indicate
gross motor delay. Refer to Table 1 for the standardized examinations’ findings.

Table 1: Test Findings from Initial Evaluation Using Justin’s Chronological Age of 24
Months.

<table>
<thead>
<tr>
<th>Test</th>
<th>Subtest</th>
<th>Score</th>
<th>Average Score*</th>
</tr>
</thead>
<tbody>
<tr>
<td>BSID-II</td>
<td>Mental Development</td>
<td>104</td>
<td>85-114</td>
</tr>
<tr>
<td>DAY-C</td>
<td>Social-Emotional</td>
<td>100</td>
<td>90-100</td>
</tr>
<tr>
<td>BSID-II</td>
<td>Psychomotor Development</td>
<td>&lt;70</td>
<td>85-114</td>
</tr>
</tbody>
</table>

*Average score for children the same age at the patient

A core evaluation was conducted on Justin. A core evaluation is a
comprehensive evaluation of the patient to evaluate each of the following domains:
physical development, cognitive development, communication development, social or
emotional development and adaptive development. During the core evaluation Justin sat
on the floor with his left foot under his bottom and his right lower extremity positioned to
the right of his body with his knee and ankle bent in a modified W-sit. His medial right
thigh, lower leg, and foot rested on the floor. Justin was able to maintain this sitting
position without using his upper extremities for support or balance. From sitting Justin
was able to transition easily onto his hands and knees to crawl. He was able to crawl
quickly to negotiate his home. Justin crawled with his right hand fisted and weight
bearing on the knuckles of his right hand. This position is thought to be a compensatory
movement pattern to clear his right lower extremity when bringing it forward to crawl.
Justin also demonstrated the ability to walk on his knees. He was able to pull to stand at
furniture by lifting himself onto his left lower extremity and lower himself down with good control onto his right knee using a wide half-kneel pattern. When weight-bearing Justin maintained bilaterally flexed knees (right knee flexion greater than left knee flexion) and externally rotated his right lower extremity secondary to the abnormal alignment of the bones in his lower extremities associated with NPS. Justin’s mother reported that he is able to cruise along the couch in either direction. Justin’s ability to cruise was not observed. Justin attempted to rise to standing from a quadruped position by transitioning onto his feet, weight shifting backwards over his lower extremities and then rising upright. He was able to transition into a semi-standing position briefly with his knee bent until his right knee buckled forcing him to fall to the floor onto his knees. Walking without assistance was not assessed to avoid placing stress on Justin’s knees.

Justin’s fine motor skills were assessed during the core evaluation. He used his eyes and bilateral hands to explore his environment and perform tasks with various items that were given to him. He was able to pick up and hold items his palm and out away from his palm with his thumb and fingers depending on the size of the item. Justin was able picked up 10 pellets using a pincer grip and place them into a container within 60 seconds. He was able to pick up a pencil with the pads of his right thumb and finger. He scribbled with the pencil in his right hand and was able to hold the paper in place with his left hand. Justin demonstrated the ability to stack blocks with accuracy and controlled release of one block on top of the other.

Evaluation

Justin demonstrated gross motor delays. He was unable to stand or ambulate independently. Justin demonstrated decreased tolerance to lower extremity weight
bearing. He was unable to pull to stand from sitting. He demonstrated decreased upper and lower extremity strength during transitional movements. His fine motor skills were age appropriate.

**Diagnosis**

Justin presents with impaired motor function and sensory integrity. The practice pattern appropriate for this patient is Neuromuscular Practice Pattern 5B: Impaired Neuromotor Development. The most appropriate ICD-9 code is 754: congenital musculoskeletal deformities.18

**Prognosis**

Justin is a good candidate for physical therapy because he is young and motivated with good family support. It is predicted that he will demonstrate the optimal highest level of functioning at home and in school with gradual improvements in strength, balance, and functional mobility.

**Plan of Care**

Interventions for physical therapy include patient/family education, home exercise plan, stretching, strengthening, balance training, gait training, and aquatic therapy. Justin was seen 2-3 times a week for 30 minute sessions until he demonstrated safe and independent functional mobility, which would allow him to negotiate in his school and community environment.

**Physical Therapy Goals:**

Annual Goal: Justin will be able to negotiate within his school, home and community independently, safely, and efficiently.
Objectives:

1. Justin will be able to walk independently 50 feet with good balance and control on even and uneven surfaces while using an age appropriate gait pattern.

2. Justin will be able to run 20 feet on even and uneven surfaces independently with good balance and control.

3. Justin will be able to ascend and descend a flight of stairs with the aid of a rail using an alternating pattern independently with good balance and control.

4. Justin will be able to transition from a sitting position on the floor to a standing position through an age appropriate ½ kneel position independently.

Annual Goal: Justin will be able to participate in age appropriate games with his peers.

1. Justin will be able to jump forward with 2 feet 12 inches independently with good balance.

2. Justin will be able to stand on one foot (right and left) for 3 seconds with abducted upper extremities without losing his balance.

3. Justin will be able to kick a playground size ball 10 feet independently while maintaining his balance with less than 45 degrees deviation 75% of the time.

4. Justin will be able to stop rolling a playground size ball with either his right or left foot without losing his balance 3 out of 4 trials.

Interventions

Justin’s aquatic therapy program was followed for 12 months with a re-evaluation after 6 months of intervention and at 18 months after interventions began. The aquatic interventions were temporary on hold twice throughout this case report. In October 2005,
Justin had an orthopedic re-alignment of his right lower extremity; he was unable to participate in an aquatic program for a month while he had a cast on. In April 2006, Justin had an orthopedic re-alignment of his left lower extremity and was unable to participate in aquatics for another month. During both time periods, Justin continued to receive physical therapy on land.

Initiation of an aquatic program began with relaxation techniques in the pool. The physical therapist would begin treatment by applying rhythmical movement at the obliques and trunk to relax the pelvic and hips. The physical therapist worked on weight bearing by giving moderate assistance at the gluteals and obliques to assist standing and lateral weight shifting. Justin performed toe touch weight bearing to improve lower extremity tolerance to weight bearing. Justin then performed a prone swim with moderate assistance at the obliques and gluteals and moderate verbal cueing to promote alternating kicking to increase strength and range of motion. These interventions were performed once a week in the pool for 6 months. Assistance level gradually decreased as the patient’s strength and range of motion improved.

After 6 months Justin became more tolerable of weight bearing with minimal assistance was needed. A walking program was added to his aquatic therapy program. Justin needed moderate assistance at his gluteals and quadriceps to promote lateral weight shifting. To increase Justin’s upper extremity strength pull-ups at the end of the pool were added to the program. Justin needed minimal assistance at his trunk to perform pull-ups. As his upper extremity strength increased, less assistance was given. The walking program and pull-ups were added to the exercises Justin was already performing. Justin had surgery in April 2006 and aquatic therapy was temporarily discontinued.
In May 2006, Justin was able to begin aquatic therapy again. The aquatic program focused on walking and weight shifting over his right lower extremity. Stair training was added to the aquatic program. Justin received moderate assistance at his quadriceps and gluteals as he worked on stepping up and step from a 3-inch step in the water. Assistance decreased as Justin was able to tolerate increased weight bearing on right lower extremity. Justin continued to work on prone swimming without assistance and minimal verbal cues to promote alternate kicking and performed wall pull-ups independently.

In July 2006, jumping and unilateral stance activities were added to Justin’s program. Justin performed two foot jumping in water with assistance from the physical therapist at his trunk to promote abdominal control. Justin was able to walk independently in the pool. The aquatic program began to focus on unilateral stance activities. Justin was able to stand on one lower extremity while shooting a basketball.

All aquatic interventions were carried over onto land. Justin received physical therapy interventions for a total of three times a week. The interventions described above would vary each session secondary to Justin’s attitude, energy level, and tolerance to activity on the day of treatment. Refer to Table 2 for a description of the aquatic therapy program performed with Justin.
Table 2: Aquatic Therapy Interventions with Progression

<table>
<thead>
<tr>
<th>Date</th>
<th>Frequency</th>
<th>Intervention</th>
<th>Purpose</th>
</tr>
</thead>
<tbody>
<tr>
<td>August 2005</td>
<td>1x per week</td>
<td>Relaxation techniques, toe touch weight bearing, prone swim</td>
<td>Increase weight acceptance, increase strength, increase range of motion</td>
</tr>
<tr>
<td>January 2006</td>
<td>1 x per week</td>
<td>Walking program, upper extremity pull-ups, prone swim</td>
<td>Improve weight bearing and ambulation, Increase upper extremity strength</td>
</tr>
<tr>
<td>May 2006</td>
<td>2x per week</td>
<td>Walking, prone swim, pull-ups, step-ups</td>
<td>Improve balance, gait and stair training</td>
</tr>
<tr>
<td>July 2006</td>
<td>2x per week</td>
<td>Step-ups, two foot jumping, unilateral stance</td>
<td>Improve balance</td>
</tr>
</tbody>
</table>

Outcomes

Justin’s gross motor skills were reassessed 6 months after his core evaluation. Justin’s primary means of mobility after 6 months of physical therapy intervention was creeping on his hands and knees with his brace on his right lower extremity. When creeping, Justin demonstrated internal rotation of his bilateral shoulders (right greater than left), right foot pronation and eversion, and left hip internal rotation. Justin was able to transition from quadruped to a sitting position by weight shifting over his left hip onto his bottom. He sat on the floor with his right lower extremity extended out in front of him with mild hip internal rotation and his left lower extremity in a “W” position. Justin was able to transition into quadruped from sitting by rolling onto his stomach, pushing up onto his hands and knees, and shifting his weight over his left hip to a quadruped position. He was able to maintain a tall kneeling position with good balance for about 4-5 minutes.
Justin was able to pull himself to stand using a support surface by positioning his right lower extremity forward with his hip externally rotated and quickly shifted his weight over his right lower extremity to unweight his left lower extremity to move it underneath him. He primarily used his upper extremities to pull to stand. Standing at a support surface, Justin presented with right lower extremity external rotation, right foot pronation and eversion, left hip internal rotation, knee flexion, and plantar flexion. He was able to stand at a support surface without leaning his body on the surface for support.

Justin was able to ascend a flight of stairs by stepping with his right foot first onto a step and shifting his weight briefly over his right lower extremity and upper extremity to step onto his left lower extremity. Justin descended a flight of stairs by lowering his right lower extremity onto a step below him and weight shifting over his right lower extremity briefly to step with his left lower extremity.

Justin was able to throw a tennis ball overhand with good shoulder rotation and direction 7-10 feet through the air. He was able to throw a tennis ball underhand with shoulder internal rotation and abduction with fair direction 5-7 feet through the air. Justin was able to catch a playground size ball with his arms by bending his elbows and trapping the ball with his chest.

Justin presented with normal forward, backward, and lateral protective reactions. He present with an incomplete, spontaneous and sustained forward righting reaction. Justin could maintain his head at midline. When pulling to a sitting position from supine, Justin presented with an initial head lag for the first ½ of the movement. He was able to tuck his chin at the last ¼ of the movement.
Justin’s gross motor skills were reassessed 6 months after his initial evaluation using the Peabody Developmental Motor Scales-2nd edition. The PDMS-2 Gross Motor Section measures gross motor skills of children from birth to 72 months. Refer to Table 3 for his scores on the Peabody at 30 months.

<table>
<thead>
<tr>
<th>Subsets</th>
<th>Age Equivalent</th>
<th>Percentile</th>
<th>Standard Scores</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reflexes</td>
<td>10 Months</td>
<td>*</td>
<td>*</td>
</tr>
<tr>
<td>Stationary</td>
<td>8 Months</td>
<td>25%</td>
<td>8</td>
</tr>
<tr>
<td>Locomotion</td>
<td>9 Months</td>
<td>&lt;1%</td>
<td>1</td>
</tr>
<tr>
<td>Object Manipulation</td>
<td>12 Months</td>
<td>1%</td>
<td>3</td>
</tr>
<tr>
<td>Average Score</td>
<td>13 Months</td>
<td>&lt;1%</td>
<td></td>
</tr>
</tbody>
</table>

* Denotes inability to score secondary to age
Gross Motor Quotient = 61

The sum of Justin’s gross motor scores places him below the 1st percentile of children in his age group. The content of each subtest is as follows: Reflexes—“Measures the child’s ability to automatically react to the environmental events”, Stationary—“Measures the child’s ability to sustain control of his or her body within its center of gravity and retain equilibrium”, Locomotion—“Measures the child’s ability to transport his or her body from one base to another”, Object Manipulation—“Measures the child’s ability to throw, catch, and kick balls”. 11
Concurrent validity between the BSID-II and the PDMS-2 are high. The correlation between the fine motor measurements is 0.87 and 0.83 for the gross motor measurements. Over the first 6 months of receiving physical therapy services Justin demonstrated an increase in protective reactions, righting reactions, and upper extremity strength as evidence by increase in ability to transition in and out of positions. He continues to demonstrate gross motor delays and decrease in strength and muscle endurance in his abdominal muscles and lower extremities.

Justin’s most recent evaluation was conducted in January 2007 using the PDMS-II. He demonstrates improvements in all areas of development. Currently Justin’s primary means of mobility is ambulating independently with moderate gait deviations and decreased balance. Justin walks with a heel to toe gait pattern with a slightly increased base of support secondary to decreased ability to weight shift laterally. He is able to transition from standing to squatting independently with a wide base of support and right lateral weight shift. He is able to maintain a squatting position for a brief period of time before falling to his right side or backwards onto his bottom.

Justin is able to ascend and descend one flight of stairs using a marking pattern with the support of one rail. He is attempting to jump with 2 feet at this time. He is able to jump by leading with his left foot with fair balance. Justin is able to throw a tennis ball overhand and underhand with fair direction. He is able to kick a soccer ball with fair balance a distance of 5-10 feet. He has demonstrated an increase in self-confidence, protective reactions, righting reactions, upper extremity strength, lower extremity strength, balance, and muscle control. Justin continues to demonstrate significant delay
in locomotor skills and a significant decrease in abdominal and lower extremity strength and muscle endurance. Refer to Table 4 for Justin’s scores on the Peabody at 42 months.

Table 4: Justin’s Score on the Gross Motor Section of PDMS-2 using his Chronological age of 42 Months.

<table>
<thead>
<tr>
<th>Subsets</th>
<th>Age Equivalent</th>
<th>Percentile</th>
<th>Standard Scores</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reflexes</td>
<td>*</td>
<td>*</td>
<td>*</td>
</tr>
<tr>
<td>Stationary</td>
<td>35 Months</td>
<td>16%</td>
<td>7</td>
</tr>
<tr>
<td>Locomotion</td>
<td>22 Months</td>
<td>2%</td>
<td>4</td>
</tr>
<tr>
<td>Object Manipulation</td>
<td>36 Months</td>
<td>25%</td>
<td>8</td>
</tr>
<tr>
<td>Average Score</td>
<td>31 Months</td>
<td>5%</td>
<td></td>
</tr>
</tbody>
</table>

* Denotes inability to score secondary to age
Gross Motor Quotient = 76

At 30 months of age Justin scored below the 1st percentile of children in his age group, at 42 months of age Justin scored below the 5th percentile of children in his age group.

At the time of Justin’s evaluation in January 2007 he had not met any of his physical therapy goals, however he is not able to walk independently with fair balance, he can negotiate stairs with a step to pattern, and he is able to maintain unilateral stance for 2-3 seconds with fair balance. Justin is currently continuing physical therapy to meet his goals.

Discussion

NPS has not been examined well in the literature. This case report gives information on the disease and aquatic interventions that can be performed with this
population of patients. The patient demonstrated improvement with activities of daily living and gross motor skills. Previous research has suggested that an aquatic therapy program can help improve strength, balance, coordination, gait and activities of daily living. The patient in this case report underwent 2 surgeries while receiving physical therapy and he was receiving physical therapy on land as well. Therefore it is difficult to state that aquatic therapy alone would be beneficial for this patient. In the future more research could be targeted toward aquatic therapy alone. During the course of therapy for this patient, the interventions varied from session to session depending on the patient’s attitude that day. More consistent sessions would allow researchers to evaluate which aquatic exercises may be more beneficial to the patient. Evaluation for this case report consisted of standardized test scores and professional judgment. Strength and range of motion testing may have been appropriate to compare results over time.

Overall the patient showed improvement in all areas of development. He is currently receiving physical therapy. The patient received 2 aquatic sessions a week and one land session. He continues to demonstrate gross motor delays when compared to children of the same age, however when compared to himself he has demonstrated significant improvement.

It is suggested that future research should focus on aquatic therapy without a land program, compared to a land program alone. This would investigate the benefits of an aquatic program in comparison to a land program to see which may provide a better intervention for these patients. It would be beneficial to look at more long-term effects of therapy for patients with NPS.
References:


March 13, 2007

Janelle L. Cassant
113 Colleen Road Apt 7
Troy, NY 12180

IRB PROPOSAL # 080-06
Reviewer: Dr. Samuel W. Hill, PhD

Dear Ms. Cassant:

I have completed the review of your project entitled “Balance Training and pool therapy for a child with Nail Patella Syndrome” and your project had been approved. Please disregard any prior correspondence regarding a withdrawal. This was sent in error. Good luck with your research.

Please let me know if you have any questions. Please refer to your IRB Proposal number whenever corresponding with us whether by mail or in person.

Sincerely,

[Signature]
Samuel W. Hill, PhD
Chair, IRB

SWH/rd

Cc: Dr. Laura Z. Gras