NAIL-PATELLA SYNDROME: DIAGNOSIS AND MANAGEMENT IN A CHIROPRACTIC SETTING– A CASE REPORT

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ABSTRACT

Objective: To describe the conservative management of a 6.5-year-old with chronic knee pain

Clinical Features: A 6.5-year-old boy had consistent right knee pain primarily at night with no known mechanism of injury. He was evaluated using history, orthopedic and neurologic examination, and x-ray. He was 3rd-percentile, and had frequent urinary tract infections, headaches, visual disturbances, and constipation. His fingernails and toenails appeared normal. Overall knee function was normal. The lateral femoral and tibial condyles were prominent and tender. X-rays revealed agenesis of the patella. Family history revealed no known members with this condition. Additional family history included glaucoma, elbow joint dislocation and kidney health disorders.

Intervention and Outcome: Home exercise recommendation to address continued biomechanical change in the knees was initiated. He responded to chiropractic intervention and age-appropriate exercises, with a reported decrease in pain and dysfunction. We monitored him at each treatment visit for concurrent health issues. We also recommended referral for co-management of his visual disturbance and urinary tract infection.

Conclusion: Early detection from portal-of-entry physicians may help improve patient health outcomes when appropriate diagnosis and referral is used. *(J Contemporary Chiropr 2022;5:187-189)*

Key Indexing Terms: Nail-Patella Syndrome; Chiropractic; Exercise

INTRODUCTION

Nail-patella syndrome (NPS) has a classic clinical tetrad of changes in the nails, knees, and elbows, along with the presence of iliac horns. Patients may have a variety of symptoms, including musculoskeletal pain, knee dislocation, decreased elbow range of motion and abnormal nail formation. There is a varying severity of NPS. Patients may also come in asymptomatic, with an incidental finding on x-ray. Nail changes are the most constant feature of NPS. (1) NPS can also bring other complications, such as early-onset glaucoma, kidney disorders, and major joint malformations. NPS is genetic and may begin in the individual's nails at birth, but nail changes are not always present. The age at diagnosis also varies. NPS has traditionally been under-reported since the range of symptoms can range from asymptomatic to needing organ replacement.

Genetic links have been found for the development of NPS, but genetic research remains in its infancy. Also, even if the genetic markers are present, the phenotype will express differently in patients, making the need to look for markers difficult since the diagnosis of NPS will not be the apparent choice. (1)

Diagnosing those with milder symptoms can be difficult with little information. It is important to try to rule out or in as many differential diagnoses as possible. For example, a pediatric patient with frequent bedwetting and NPS would need to be examined for urinary tract infections sooner, with a referral to a nephrologist to check kidney function. Also, the standard eye exam for a pediatric well-child vision check does not include a glaucoma test, which for an NPS patient would be relevant. Chiropractors are in a great position to aid in the diagnosis and management of this condition since they focus on whole-body dysfunction and the root cause of patient symptomatology. Chiropractors may also provide lifestyle advice to patients with NPS and have can referg to other specialties when major symptoms arise.

CASE REPORT

A 6.5-year-old boy was brought for care for chronic headahces and chronic right posterior knee pain that woke him at night. His family history was positive for glaucoma on the father's side. The child's father had trouble extending his left arm at the elbow. The child had a history of urinary infections whose cause could not be

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identified. No night sweats were reported. He had visual disturbances that included being hurt by seeing the colors yellow and grey. His diet was nutritionally poor, with little water intake and a history of constipation. His parents mentioned possible undiagnosed ADHD, which was shared with his 5-year-old brother. He was on no current medications and had a history of chiropractic care.

His temperature was 98°F, blood pressure 99/45, his height was 49 inches and he weighed 46.3lbs. His BMI was 13.56 (3.28 percentile for his age). He was tall and slender. The area behind his right knee was tender to touch and had a palpatory nodule located on the posterior lateral knee joint. He had a positive valgus stress test on his right knee, normal range of motion (ROM) at the knee joint, and normal upper and lower limbmuscle strength (5/5) and reflexes (2+). His fingernails and toenails appeared normal on inspection. Eye and cranial nerve tests were normal.

X-ray imaging was discussed with parents to aid in diagnosis. With his presenting symptoms, our differential diagnosis included osteochondroma, osteoid osteoma and normal growing pains. The imaging would help by visualizing the painful nodule and guiding management. Imaging showed an aplastic patella on the right. This finding, with the family history of early onset glaucoma, ADHD, and father's problematic elbow joint restrictions led us to the diagnosis of NPS.

Initial Therapeutic Intervention

With the knowledge of the weakness of the vastus medialis muscle and joint pain involved here, our focus was to recommend physiotherapy to address strengthening the quadriceps muscle. We taught the child (and his parents) proper sitting and standing posture to keep stability at the knee joint. When teaching this, we found that he had the habit of "W-sitting" (sitting on the floor with knees bent and hips internally rotated) and we therefore asked the parents to help him to sit normally. We also incorporated good jumping and landing techniques.

He received weekly chiropractic adjustments. We used Activator Technique to adjust misaligned vertebral segments of the sacrum, thoracic and cervical spine, and his right knee joint. By his 3rd visit, his range of motion improved and his pain resolved. The pain in the his knee decreased and his parents reported less complaints at home after beginning the chiropractic and home-exercise treatment. Re-evaluations were scheduled for every 6 months to check for stability and how his original symptoms were progressing.

Follow Up Visits & Assessments

Due to the patients' age and his diagnosis of NPS, follow-up x-rays were not taken. Advice was given to his parents to have regular check-ups for glaucoma and kidney function. His parents verbally confirmed, over the treatment course, that he was sitting cross-legged and working on correct posture. Parents reported child began sleeping through the night and the child denied having any further pain.

DISCUSSION

Diagnostic criteria have not been published for NPS. The parents here became well-informed on how to help their son. Prior to diagnosis, the ADHD symptoms the child displayed were taken as "boys-being-boys," but with studies from López-Arvizu (2) that reported a higher prevalence of ADHD in NPS patients, the parents were able to recognize a cause. Given that the child was underweight continuing to track his skeletal development is helpful; Towers (3) reported that individuals with NPS have a lower bone-mineral density than the general population. Early intervention has also seen to be beneficial in a study that looked at the social and emotional impact NPS can have on individuals (4) as well as the measures were taken for early detection of eye and kidney issue development. The parents are now aware of NPS symptom manifestation and have a better understanding of their child's condition.

Limitations

This case report only demonstrated the mild symptoms present with NPS. There was no genetic testing to support the diagnosis. Since NPS is a genetic disorder with an array of possible presentations, this report shows only management and referral to guide patients in monitoring possible disease processes, as well as steps to take for preventive care. We are not able to provide long-term reports needed to fully see the outcome of the early intervention and diagnosis of this disease.

CONCLUSION

Early detection from a portal-of-entry physician was a valuable tool in helping a patient and his family manage NPS. It demonstrates how heightened awareness for the presentation of possible complications can be lessened when caught early.

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