Undiagnosed Nail Patella Syndrome in an Elderly Chiropractic Patient: A Case Report

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ABSTRACT

Background

Nail-Patella Syndrome is a rare autosomal dominant condition with high penetrance and varied clinical expression, affecting fewer than 20 in 1 million persons. The most common features include nail dysplasia, hypoplastic or absent patella, renal anomalies, and ophthalmological disease.

Case Presentation

A 67-year-old female presented to a chiropractic clinic for symptoms of hip pain with radiation to the knee. The patient's history was remarkable for glaucoma, retinal surgeries, and current cigarette smoking. Physical examination results were remarkable for hypertension, delayed upper and lower extremity capillary refill, cervical and lumbar muscle spasms, and reduced cervical ranges of motion. Radiographic examination revealed osteopenia, diffuse idiopathic skeletal hyperostosis, osteoarthrosis, and iliac horns (bony exostoses) from the posterior ilia.

Results

The patient was diagnosed with Nail-Patella Syndrome, from the radiographic and clinical findings, and with intersegmental dysfunction, hypermyotonicity, osteoarthrosis, and osteopenia. Treatment included chiropractic manipulative therapy, postural exercises,

electrical muscle stimulation, and cryotherapy. The patient's musculoskeletal symptoms fully resolved within 3 weeks. She was referred to nephrology and ophthalmology for evaluation of associated complications of Nail-Patella Syndrome.

Conclusion

An elderly patient with musculoskeletal symptoms experienced pain relief with conservative treatment and was diagnosed with a rare inherited disease not previously recognized by her other physicians or by family members with similar clinical features. The discovery led to specialist consultations in ophthalmology and nephrology for management of possible associated complications of Nail-Patella Syndrome.

Key Words: Nail-Patella Syndrome, Fong Disease, Iliac horns

INTRODUCTION

The atypical patellae and dysplastic nails of the rare hereditary disease known since the 1950s as Nail-Patella Syndrome (NPS) were first described in scientific literature in 1897.[1] Subsequent authors described additional associated features, resulting in several eponymous and descriptive names for NPS, including Turner syndrome, Fong disease, arthro-onychodysplasia, and hereditary osteo-onycho-dysplasia (HOOD).[1]

Nail-Patella Syndrome is a rare hereditary condition of both ectodermal and mesodermal tissues,[2] occurring in approximately 4 to 20 persons per 1 million live births.[3,4] NPS is an autosomal dominant inherited condition with high penetrance and variable expressivity.[4] The condition's inconsistent clinical features may result in delayed diagnoses,[3] even amongst multiple impacted family members, as was the case in the patient described.

In 1998, the NPS gene was identified as *LMX1B* on chromosome 9q.[2,5,6] Only 12% of reported NPS cases are due to a spontaneous genetic mutation.[6] The disease is typically diagnosed through a combination of clinical and radiographic features. Genetic testing may be performed if the clinical and radiographic evaluation is equivocal.

Clinical findings of NPS may include:

- **Nail/digit anomalies, pathognomonic** (95.1%)[1,6]: anonychia, hypoplastic (micronychia),[6] or dystrophic fingernails, with the thumb most frequently involved; triangular-shaped lunulae; decreased creases over the distal interphalangeal (DIP) joints; the toenails are less commonly affected
- **Patellar anomalies, pathognomonic (**92.7%)[6]: absent or hypoplastic patellae; superior and lateral patellar dislocations; early osteoarthritis
- **Iliac horns, pathognomonic** (80%)[1,6]: asymptomatic, possibly palpable, posterolateral iliac exostoses at the origin of the gluteus medius, present in approximately 70% of patients, but considered pathognomonic for NPS when identified; iliac horns do not affect the patient's gait or gluteus medius strength[7,8]

- **Elbow anomalies** (92.5%)[6]: reduced flexion, supination, and pronation ranges of motion; possible hypoplastic and posteriorly displaced radius; antecubital webbing (pterygia) of the elbow skin may be present [7]
- **Renal involvement** (26-60%)[6]: proteinuria with or without hematuria, with progression to end-stage renal disease in approximately 5-30%[6,8]; hypertension
- **Ophthalmological disease** (10-16.7%)[1,4]: open-angle glaucoma with optic nerve and visual field damage (peripheral field deficits); ocular hypertension; Lester sign, a cloverleaf hyperpigmented ring in the iris
- **Lower extremity anomalies**: talipes equinovarus, equinovalgus, absence of the fibula[6]

CASE PRESENTATION

A 67-year-old Caucasian female sought chiropractic care for constant sharp left iliofemoral pain with radiculopathy to the left knee. The pain began approximately one-month prior after she lifted several heavy boxes. Her symptoms were worsened by the activities of climbing stairs, walking, crossing her legs, lying on her side, getting in and out of the shower and car, and putting on her shoes. Sitting in a recliner and taking naproxen sodium lessened her pain. Additionally, her pain was worse in the mornings and afternoons and improved in the evening hours.

Examination

The patient was cooperative and attentive during the examination, oriented to person, place, and time, but with pallor, a drawn facial expression, and a forward antalgic posture.

Vital sign examination results were as follows: radial pulse of 64 beats per minute, respiration of 14 breaths per minute, temperature of 98.5 degrees F, and right brachial blood pressure of 138/72 mmHg. The patient's heart sounds were clear and distinct without murmurs, her bowel sounds were present, and her abdomen was soft and non-tender. Bruits of the carotid and subclavian arteries were not detected. Upper and lower extremity capillary refill was delayed with returns greater than 4 seconds. Brachioradialis, biceps, triceps, patellar, and ankle reflexes were brisk and graded at 2+. Upper and lower extremity muscle strength and tone were all 5/5 and light touch and pinprick sensation were intact.

Cervical lateral flexion and rotation were reduced with muscle spasms present. Lumbar extension was reduced, and pain and muscle spasm were noted. Several provocative orthopedic examinations resulted in increased pain lower lumbar pain, including Kemp's test, Seated Straight Leg Raise, Supine Straight Leg Raise, and Fabre-Patrick. Valsalva test did not increase pain. The patient revealed she is a smoker, has glaucoma, and had multiple eye surgeries including selective laser trabeculoplasty. Her mother died at age 67 and her maternal grandfather died at age 91, both due to colon cancer. No additional information regarding her family health history was reported during the initial interview.

Imaging

Radiographic evaluation of the lumbar spine was performed, revealing osteopenia, degenerative disc disease, zygapophyseal facet arthrosis without spondylolisthesis, Diffuse Idiopathic Skeletal Hyperostosis (DISH), and arterial calcifications without aneurysm of the abdominal aorta and iliac arteries. Uncommon and unexpected radiographic findings of large, triangular, bony projections from the bilateral posterolateral ilia were identified. (**Figures 1A and 1B**).

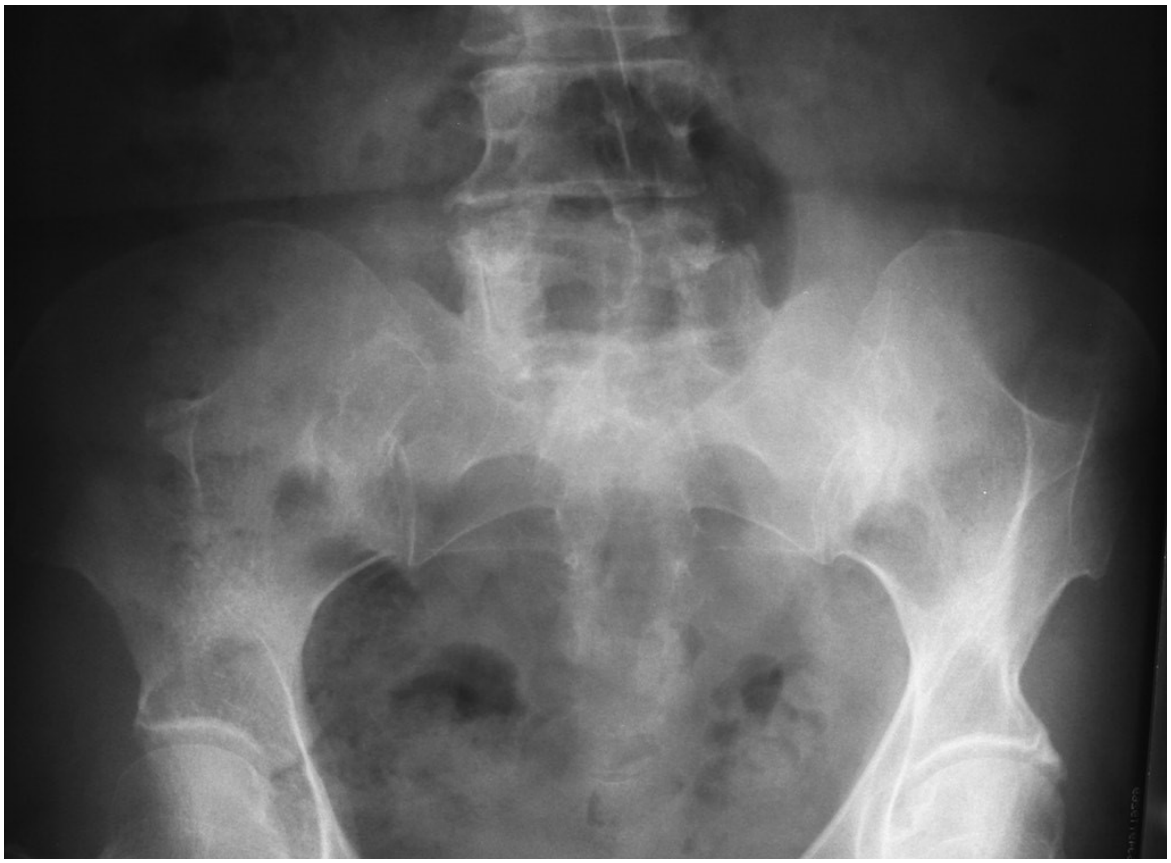

Figure 1A: Bilateral posterolateral iliac exostoses.

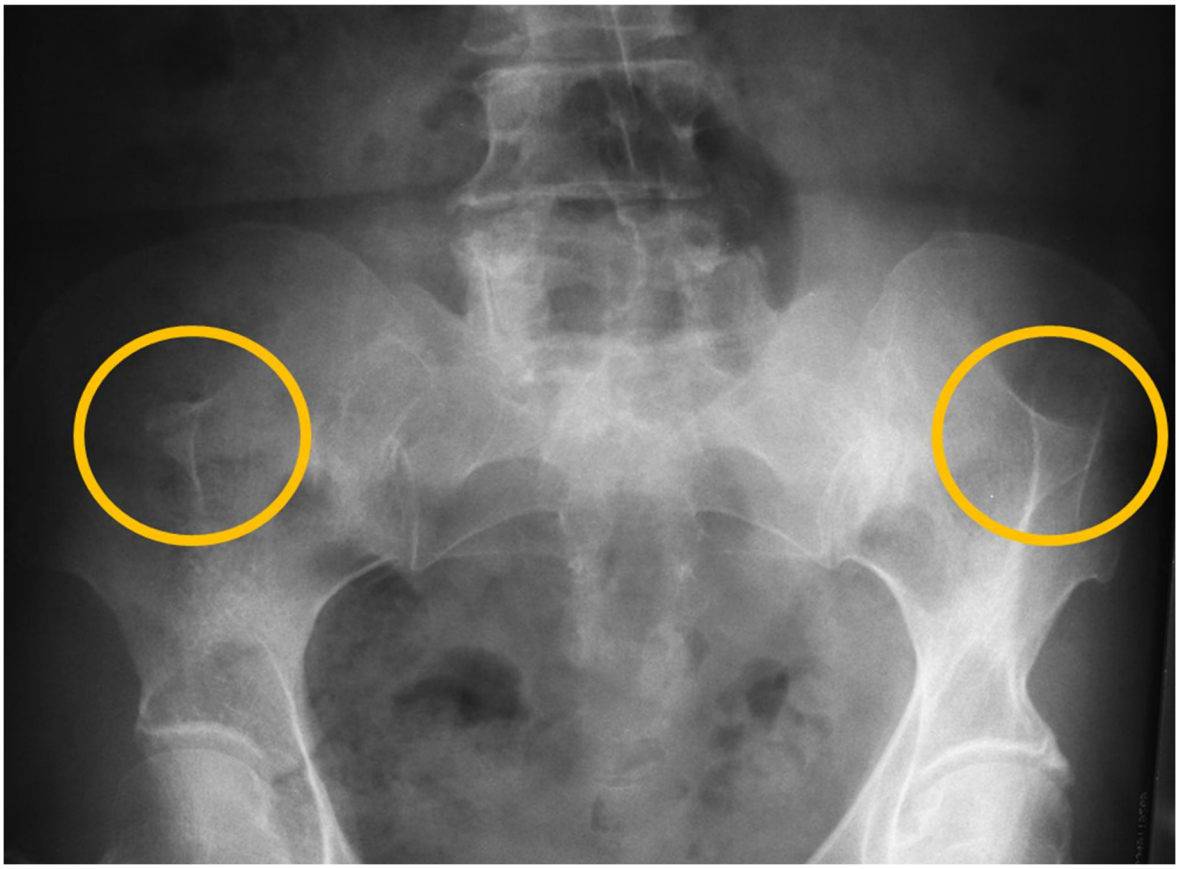

Figure 1B: Bilateral posterolateral iliac exostoses, annotated.

Iliac-based exostoses ("iliac horns") are a pathognomonic finding of Nail-Patella Syndrome (NPS), also known as Hereditary Osteo-Onychodysplasia Disease (HOOD), Turner-Keiser syndrome, and Fong disease. The patient was informed of the diagnosis of NPS, and additional evaluations of the small patellae and hypermobile elbows not identified during the initial examination were performed. The patient subsequently revealed that several family members had "nail problems", including her mother and her son, but no family member had ever received a diagnosis related to the unusual appearance of their nails. (**Figure 2**).

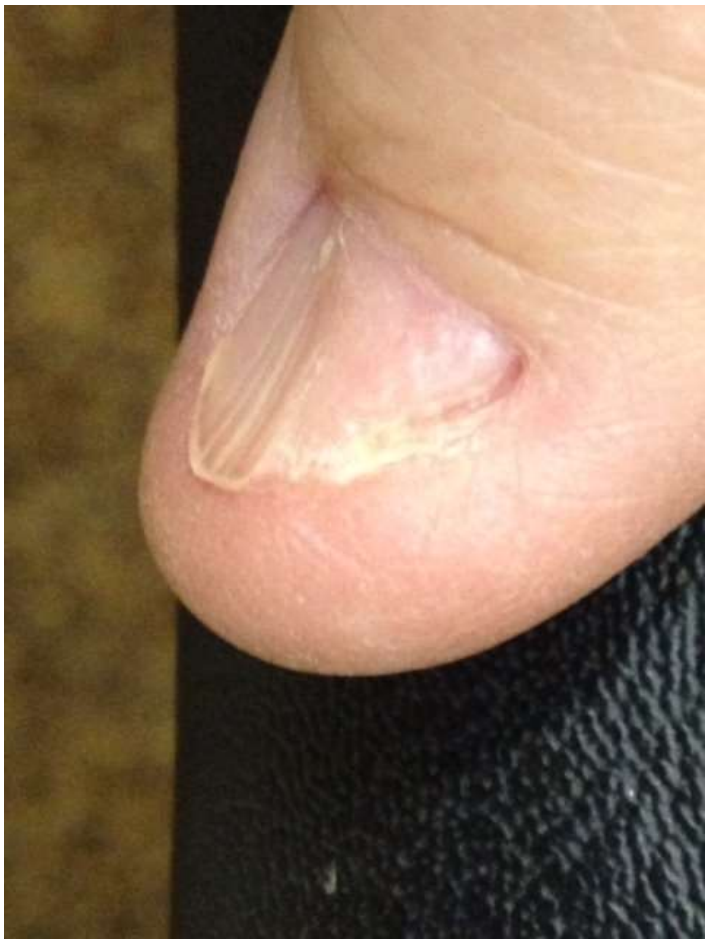

Figure 2: Dysplasia of the patient's thumbnail

Additional diagnoses included osteoarthritis, intersegmental fixation and dysfunction, and hypermyotonicity of the trunk and pelvic musculature. The patient was treated with a combination of chiropractic manipulative therapy, postural exercises, electrical muscle stimulation, and cryotherapy. During the first three weeks of treatment, the patient's pain levels decreased, and muscle spasms were alleviated. She was referred to ophthalmology and nephrology for assessment of complications of the NPS.

DISCUSSION

There is no cure for NPS. The offspring of one parent with NPS have a 50% risk of inheriting this disease.[8] Genetic assessments, prenatal counseling, early diagnostic exams, and surveillance of patients and affected family members are integral to diagnosing the varied expressions of NPS, including the orthopedic, ophthalmological, and renal complications of the disease.[8] Non-steroidal anti-inflammatory medications (as was utilized by the patient in this case report for analgesia) should be avoided due to the detrimental impact on renal function.[9]

CONCLUSION

This is an unusual case of an elderly patient with undiagnosed Nail-Patella Syndrome, despite lifelong signs present in her and multiple family members. This case report emphasizes the importance of early identification of symptoms and signs of NPS to address and reduce the musculoskeletal, ophthalmological, and renal complications associated with this disease.

LIMITATIONS

This is a single-patient case report. Results may not be generalizable to other individuals with similar conditions.

CONSENT

The subject of this case report is deceased, thus she could neither grant nor withhold consent to the use of her de-identified personal information.

COMPETING INTERESTS

The authors declare no competing interests.

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