Knee Arthrodesis in Navajo Familial Neurogenic Arthropathy

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Abstract

A 16-year-old Native American male with Navajo familial neurogenic arthropathy presenting with a Charcot joint of his right knee was treated with knee arthrodesis and is reviewed 1 year post surgery. Despite complications including peroneal nerve palsy, knee arthrodesis has restored the ability to ambulate. Both Navajo arthropathy and indications for knee arthrodesis in children are rare. As the Navajo population disperses into the greater community, health professionals should be aware of Navajo familial neurogenic arthropathy and treatment options. Traditional Navajo beliefs require a cautious approach when discussing prognosis and possible adverse outcomes.

Introduction

Navajo familial neurogenic arthropathy (NFNA) is a rare form of hereditary sensory autonomic neuropathy found within the Navajo population. It usually manifests during the first two decades of life with unrecognized fractures, Charcot's joints resulting from sensory deficits to deep pain, hypohidrosis and heat intolerance.¹ The orthopedic manifestations secondary to NFNA has been investigated,² however, evidence regarding treatment outcomes and prognosis is limited.

Case Report

An 8-year-old Navajo boy presented for pediatric orthopaedic consultation with painless right knee effusion several weeks after injuring his knee while wrestling with his father. The patient had no significant medical history and no family history of musculoskeletal problems. He admitted to sweating little and frequently became overheated. Physical examination revealed a knee effusion, a posterior drawer sign, a positive Lachman sign of approximately 8 millimeters, and a grade I pivot shift. Additionally, the patient exhibited dry, thickened palmar and plantar skin, absent muscle stretch reflexes, and lack of pinprick sensation. Radiographs revealed a large joint effusion about the knee with bony sclerosis and destruction involving the lateral femoral condyle and patella (Figure 1). These findings, coupled with his lack of pain, suggested Charcot arthropathy. Diagnostic arthroscopy confirmed a large osteochondral defect in the lateral femoral condyle, absence of the anterior cruciate ligament, and patellar erosion of uncertain duration.

Neurologic consultation was obtained. An MRI of brain and spinal cord revealed a Chiari I malformation. The EMG/NCS results included an absent sural sensory response, a reduced amplitude median sensory response, and an absent galvanic skin response on the palm and dorsum of the foot consistent with autonomic neuropathy. The patient was diagnosed with Navajo familial neurogenic arthropathy based on clinical presentation and nerve conduction.

Initial treatment included brace stabilization with a brief discussion of eventual surgical options. Treatment options were introduced cautiously, because in the Navajo culture a discussion of possible bad outcomes may set into motion the circumstances resulting in that bad outcome, and is considered to bring bad luck.³ Other cultural considerations include the fact that allograft bone or other cadaver tissue grafts are unacceptable to the traditional Navajo, who has a strong death taboo.⁴ Given the progressive nature of neurogenic arthropathy, the only viable surgical options would have been knee arthrodesis or amputation, neither of which was appealing to the family.

The patient was followed periodically from age 8 to age 15, and surgical options were gently explored at each visit. Despite attempts at stabilization using knee-ankle-foot orthoses of various types, the right knee progressively deteriorated. At age 16, the knee was the size of a basketball, and instability was so great that ambulation was impossible. Radiographs revealed massive joint destruction and limb length discrepancy secondary to lateral subluxation of the tibia on the femur (Figure 2).

The grotesque appearance of the knee and inability to walk prompted the family to consider surgery. They preferred an attempt at arthrodesis rather than amputation. Preoperative discussion avoided using direct negative personification such as, "your child could lose

Figure 1. Anteroposterior and lateral radiographs of right knee at age 8.

Figure 2. Standing radiograph at age 15, preoperatively.

his leg.” Instead, third person references were used, such as, “in some children, a knee fusion does not work and the leg must be amputated.”

A knee fusion was performed using a custom intramedullary interlocking nail extending from the trochanter to the distal tibia (Figures 3 and 4). Postoperative complications included a 3-cm skin necrosis on the lateral aspect of the right knee, orthostatic intolerance during recovery due to supraventricular tachycardia, and a peroneal nerve palsy, with numbness to touch and foot drop. Postoperative limb length discrepancy was 6.5 cm.

At 12 months postoperatively, the patient and his family report being very satisfied with the surgical outcome despite persistent

peroneal nerve palsy and foot drop. He has no pain and ambulates easily: the limb length discrepancy is not bothersome to him. With knee stiffness and foot drop (he declines orthotic use), the limb length discrepancy facilitates swing-through during gait.

Discussion

Navajo familial neurogenic arthropathy (NFNA) was described by Johnsen in 1993.¹ Found in the Southwest Navajo population, presentation is typically within the first two decades life with unrecognized fractures, Charcot’s joints, hypohidrosis and heat intolerance. Associated sensory deficits can vary from no notable deficits to abnormal sensation to deep pain, poor temperature discrimination, and corneal insensitivity. Reflexes usually remain intact, muscle strength is normal, and electromyography and nerve conduction velocities are within normal limits. Sural nerve biopsy reveals reduction in small myelinated and unmyelinated nerve fibers. Family history suggests an autosomal recessive inheritance pattern.

Differential diagnosis includes Navajo neuropathy (NN),⁵ and hereditary sensory autonomic neuropathy (HSAN) type IV.^{6,7} NFNA is differentiated from these other neuropathies by absences of the

Figure 3. Standing radiograph one year postoperative.

Figure 4. Lateral radiograph one year postoperative.

following: hypotonia, slowed nerve conduction, liver dysfunction, self-mutilating behavior, and mental retardation.⁵⁻⁷ HSAN-IV has been associated with mutations on the TRKA gene,^{8,9} while genetic associations with NFNA have not been identified.

Woiczik and D’Astous at Utah Shriners Hospital first described the orthopaedic manifestations of Navajo familial neurogenic arthropathy with a review of two cases in children.² In the second decade of life, these children exhibited Charcot-type arthropathy, heat intolerance, and anhidrosis. Surgical interventions (osteotomy and hemiepiphysiodesis) had suboptimal outcomes and many complications occurred. Physical function declined, with additional deformities of the appendicular and axial bones. Johnsen¹ mentioned arthrodesis as an option in these patients, yet to date, outcomes have not been reported.

Navajo cultural considerations make discussion of surgical treatment options with the patient and family challenging. One should avoid personalizing negative information when talking with patient and family regarding prognosis and surgical risks.³ The Navajo by nature are reticent, and questioning those perceived as persons of respect is seen as rude¹⁰; patient-centered

communication strategies include allowing ample time for any discussion and the use of “third party language” when discussing possible complications.¹

By definition, neurogenic arthropathy is a progressive disorder, and involvement may spread to other joints and to the spine. Using culturally appropriate language, patients must be made aware of the progressive nature of NFNA, and must be given reasonable expectations of surgical risks and long-term outcomes.

As the Native American population disperses into the greater community, physicians must be aware of the manifestations of NFNA. Traditional Navajo beliefs necessitate a cautious approach when discussing potential adverse outcomes and the progressive prognosis of the disorder.

Indications for knee arthrodesis in children are rare, but in this instance, the procedure avoided amputation and enabled independent ambulation, for at least the immediate future.

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