Multiple Pterygium Syndrome With Severe Knee Flexion Contracture: A Case Report

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ABSTRACT
Multiple pterygium syndrome, a subgroup of arthrogryposis multiplex congenita, is characterized by the webbing of different parts of the body. We describe a child who developed 140° flexion contractures of the popliteal fossa, which complicated mobility, skin care, and general hygiene. After seeking multiple opinions, the parents elected to proceed with through-knee amputation of both knees. At 6 month follow-up, the child had been fitted with bilateral prostheses and was able to ambulate with assistance. The findings of the current case suggest that in extreme cases of multiple pterygium syndrome, through-knee amputations may represent a predictable and functional treatment option that should be considered.

Keywords: Multiple Pterygium Syndrome, Contracture, Prosthesis and Implants, Amputation

INTRODUCTION
Multiple pterygium syndrome is a clinically heterogeneous disorder described as the pterygia (ie, webbing) of various parts of the body and associated craniofacial anomalies. This syndrome is a subgroup of arthrogryposis multiplex congenita, characterized as multiple joint contractures across different parts of the body. Other subgroups include amyoplasia, distal arthrogryposis, fetal crowding, and systemic tissue disorder. In multiple pterygium syndrome, the most commonly affected areas include the neck, axilla, antecubital, and popliteal. Patients with this syndrome can present with various features such as short stature, syndactyly and camptodactyly of the fingers, foot deformities, facial abnormalities, scoliosis, and neurologic dysfunction.1–5

Multiple pterygium syndrome is an uncommon condition with few documented cases. Usually, the syndrome is initially seen in utero with decreased fetal movement noticed in ultrasound and reported by mothers.6 It is suspected that multiple pterygium syndrome is a multifactorial condition resulting from gene defects or problems with the intrauterine environment; however, specific genetic abnormality for multiple pterygium syndrome has not been detected yet. Fetal akinesia produces maldevelopment of the joints and excessive connective tissue.2 The resulting flexion contractures present extreme challenges for patients and their providers. We describe a patient who underwent through-knee amputation of both knees to treat multiple pterygium syndrome that was diagnosed in utero.

CASE REPORT
Our patient first presented as a 3-week-old female newborn with severe pterygoid contractures since birth. Flexion contractures of both elbows and radial longitudinal deficiency (ie, radial club hand) of the right hand were diagnosed. Initially, the elbow contractures were treated with extension splinting. At that time, it was thought that functional outcome of the upper extremities was achieved.

At 3 years of age, the patient returned to the clinic. She lived at home with her mother, older brother, and maternal grandmother. She still had considerable contractures of both elbows; however, they remained functional. Unfortunately, the flexion contractures of her knees had progressed to 140° with little motion (Figures 1A and 1B). The patient’s mother reported difficulty with performing her daughter’s daily hygiene routine, concerns about the developing skin breakdown that was in the popliteal fossa of both knees, and challenges regarding wheelchair transportation. At this time, through-knee amputations were discussed. It was felt that this option would provide the best functional outcome, but the family was encouraged to seek a second opinion because of the magnitude of the treatment. Additionally, arrangements were made for the mother to meet with two different occupational
therapy specialists to ascertain whether the patient could use the proposed prosthetics owing to upper extremity limitations.

Two years later, at age 5, the patient returned to the clinic. Her mother reported that the large referral center they went to for a second opinion had agreed with our plan. The patient primarily used a wheelchair and otherwise had to crawl for mobility. Her skin breakdown continued to be an issue and no improvements had been seen. Her mother wished to proceed with through-knee amputation of both knees, with the goals of preventing further skin breakdown, improving balance while sitting, and potential ambulation with prosthetics. The orthotics and prosthetics specialist visited with the family again to discuss prosthetic options.

The surgical procedure was performed without complication, and the patient was discharged from the hospital 2 days postoperatively (Figure 2). The patient was noted to have promising healing of her surgical wounds but was thought to have developed phantom pains that were then controlled with a low dose of gabapentin.

At 5 months postoperatively, the patient had been fitted with prostheses for both knees (Figure 3). She began once-a-week sessions with a physical therapist. At 6 months postoperatively, her physical therapist reported that she was able to ambulate about 91.4 m (300 ft) with moderate assistance (Figures 4A and 4B). Overall, the mother had no concerns and was happy with the outcome.

**DISCUSSION**

Multiple pterygium syndrome is an uncommon condition for which little has been published regarding treatment and outcomes. To our knowledge, this is first report detailing 140° flexion contractures of the right and left knees. Our patient had popliteal skin breakdown, could only crawl for mobility, and had poor sitting balance and wheelchair fit. Our options were continued observation, dynamic splinting, soft-tissue releases, or amputations. Owing to the lack of published data on treating such severe contractures, our team felt that through-knee amputation of both knees would provide the most predictable outcome. Her
mother was present at all clinic visits and was intimately involved with this decision.

Other techniques have been described for treating less severe knee contractures. One technique uses a Z-plasty to lengthen the soft tissues. This option can only be used for small contractures and when the sciatic nerve and vessels are not displaced into the webbing. One patient with a 45° contracture was treated with soft-tissue releases and gradual lengthening using an Ilizarov external fixator. Although initially successful, 15° to 30° of contracture recurred postoperatively. In the current case, soft-tissue releases were not believed to provide a functional outcome. Although it was not confirmed with advanced imaging findings, the sciatic nerve and vessels commonly adhere to the pterygium.

In the current case, we had the benefit of being involved from the beginning. Early in the patient’s life, her mother had been counselled about the potential of amputations. Her mother returned to the clinic with a clear understanding of what she was agreeing to after receiving a second opinion, meeting with several therapists and prosthetics specialists, and having multiple visits with our orthopaedic team. Although not a common surgical solution, having our team ready and on the same page helped ease the transition for the family. Furthermore, the surgical procedure was successful, without any complications, and the patient continued to progress with physical therapy. At 6 months postoperatively, she was standing and walking up to 91.4 m (300 ft) with moderate assistance using knee prosthetics and handheld support at the elbow and forearm.

When carefully planned and tailored to the individual case, through-knee amputation may provide a predictable and functional solution for young patients with severe knee flexion contractures associated with multiple pterygium syndrome. In the current case, no risk of further reoccurrence has been observed and physical therapy was started immediately after the procedure. Through-knee amputation should be discussed as a surgical option in these uncommon circumstances.

REFERENCES