Distal lower extremity manifestations in spina bifida patients of the Yucatan Peninsula: A 24-year retrospective case series

by Alexandra Heidtmann, BS¹; Lahari Madulapally, BS, MA¹; Luis Rodriguez Anaya, DPM²; Daniel Cawley, DC, MS²

The Foot and Ankle Online Journal 13 (4): 8

Spina Bifida, a rare congenital disorder with an incidence of 7.85 per 10,000 births in Mexico. It results from the failed closure of the neural tube leading to the incomplete development of the neural arches. This case series is part of the Yucatan Crippled Children’s Project that began in 1996 by Charles Southerland, Doctor of Podiatric Medicine and former professor of Barry University’s School of Podiatric Medicine. All patients in this study were assessed and treated at the Red Cross hospital in the city of Merida, Yucatan, Mexico. Attendings, residents and medical students travel to the Yucatan Peninsula four times a year for a period of one week. Given that this study was built from reports of medical mission trips that occur four times a year with limited resources and time, the lack of documentation of treatment plans and follow-ups made it difficult to identify surgical procedures and assess the success of surgeries. Additionally, we did not have access to the patients birth records or their mothers medical records to accurately determine the etiology of their deformities. Based on our data, we conclude that intervention should be considered as early as possible in any flexible deformity to prevent them from becoming rigid.

Keywords: Spina Bifida, talipes equinovarus, clubfoot, adductovarus, calcaneo valgus, Ponseti, osteomyelitis

Spina Bifida, a rare congenital disorder with an incidence of 7.85 per 10,000 births in Mexico. It results from the failed closure of the neural tube leading to the incomplete development of the neural arches [1]. Spina bifida is the result of genetic and non-genetic factors that interfere with the folding and closure of the neural tube. In its most severe form, meningomyelocele, the neurons of the spinal cord are exposed to amniotic fluid resulting in neuronal death. In addition, the spinal cord and meninges protrude through the midline bony defect of the back [2].

The clinical manifestation of a meningomyelocele is dependent on the spinal level of involvement and the presence of cerebral involvement and hydrocephalus [3]. Sensory and motor impairments are commonly present below the level of the lesion causing alterations in the bowel and bladder function, muscle paresis and paralysis, and sensory loss. Impairment is classified by the level of neurosegmental involvement determined by the strength of specific muscle groups [3]. Nearly all patients with spina bifida will experience manifestations in their feet, especially those cases involving the thoracic and lumbar spinal regions [4,5]. Previous studies have reported that the most common manifestation of spina bifida in the feet are talipes equinovarus, equinus, vertical talus, calcaneal deformities, and cavovarus [4,6,7]. The aim
of this study is to analyze the incidence of various distal lower extremity manifestations and their long-term effects on spina bifida patients of the Yucatan Peninsula.

Methods

This case series is part of the Yucatan Crippled Children's Project that began in 1996 by Charles Southerland, Doctor of Podiatric Medicine and former professor of Barry University's School of Podiatric Medicine [8]. All patients in this study were assessed and treated at the Red Cross hospital in the city of Merida, Mexico. Attendings, residents and medical students travel to the Yucatan Peninsula four times a year for a period of one week.

From 1999 to 2020, we retrospectively analyzed 1,489 patients that were seen by physicians from the Yucatan Crippled Children’s Project. Among the total, we identified 25 patients, 17 male and 8 female, with history of Spina Bifida and concomitant lower extremity deformities. From the 25 patients, 15 patients had bilateral lower extremity deformities and 10 patients had unilateral deformities, leading to a total of 40 limbs. The ages ranged from 3 months old to 43 years old, with a total average age of 11.33 years. The mean age for rigid deformities was 15.4 years, while the mean age for flexible deformities was 8.8 years.

We analyzed 3 cases of patients with a history of spina bifida and lower extremity deformities according to the clinical notes collected from the Yucatan Crippled Children’s Project.

Case Presentation 1

Case 1 is a 2-year-old male who presented to the Yucatan Crippled Children's project clinic in March of 2007 with a chief complaint of difficulty ambulating. Patient's family reports past medical history of birth at 38 weeks and significant time spent in the NICU due to hydrocephalus and spina bifida. Upon initial assessment, the patient was alert and oriented and showed no additional symptoms. Patient was diagnosed with bilateral flexible clubfoot deformity, as seen in Figures 1 and 2. Considering the age and the flexibility of the deformity, the conservative Ponseti serial casting technique was performed on the patient.

The patient and his family were instructed to follow-up with the local doctor. The patient returned to the clinic in November of 2017, at 12 years old, with chief complaint of continued difficulty ambulating due to the progression of the windswept deformity (Figures 3 and 4). After assessment, the left foot was diagnosed with adductovarus and talipes equinovarus deformity. The right foot was diagnosed with forefoot adduction, midfoot abduction, and calcaneovalgus which are the three components of complex skew foot. Given the Ponseti technique applied ten years ago had failed, the deformity has worsened and progressed from flexible to rigid. The procedure consisted of application of external fixation with medial motor for gradual correction of adductovarus deformity on the left foot (Figures 5-7).
Figure 8 Bilateral flexible cavovarus deformity with ulcer on dorsolateral aspect of right foot.

Figures 9 and 10 Nine year follow-up shows rigid bilateral cavovarus deformity. The patient is confined to a wheelchair.

The patient presents to the clinic in February of 2018 for a 3 month postoperative visit after application of external fixation. It was noted that the toes were not fixated during the external fixator surgery and they developed flexion contractures within the reduction frame. The patient developed clinodactyly of all five digits of the left foot. At this date, the frame was removed and the patient began physical therapy in an attempt to reduce flexion contractures.

Case Presentation 2

Case 2 is an 8-year-old female with a past medical history of spina bifida and sensory neuropathy bilaterally. The patient presented to the clinic in July of 2011 with a chief complaint of a wound on the right foot. Upon physical exam, an open ulcer was noted on the dorsolateral aspect of the right foot along the cuboid-5th metatarsal joint (Figure 8). The wound has a beefy red base with friable granulation tissue and a circumferential macerated periwound with suspected areas of hyperkeratotic tissue. Mild hyperpigmentation and erythema is noted proximally towards the dorsum of the ankle. The patient was diagnosed with a pressure ulcer on the right foot and bilateral flexible cavovarus deformity. Ulcer was managed during the patient’s first visit prior to any surgical intervention. Upon healing of the ulcer, surgery was performed; however, the surgical technique was not recorded.

The patient was virtually contacted during Covid-19 2020 Pandemic, and sent Figures 9 and 10. The patient stated she still has insensate feet and is unable to ambulate. Deformity has progressed to rigid and she is waiting until the next Yucatan Medical Mission Trip to possibly undergo another surgery that would allow her to ambulate.

Case Presentation 3

Case 3 is a 24-year-old male with a past medical history of spina bifida, insensate feet, and chronic lymphangitis. The patient presented to the Yucatan Project Clinic in April of 2005 with a chief complaint of wounds on the right foot. Upon physical exam, ulcer on the lateral aspect of the head of the 5th metatarsal of the right foot was noted to have a 50/50 granular fibrotic base with slough in the center. The periwound consisted of hyperkeratotic tissue on the plantar aspect of the 5th metatarsophalangeal joint. Hyperpigmentation is present extending proximally on the dorsum of the foot. The second wound, located on the lateral aspect of the 5th metatarsal tuberosity of the right foot, appeared to have 75/25 fibrotic granular base with regular borders. The patient was diagnosed with active infected ulcers and bilateral cavovarus deformity (Figures 11 and 12). Initial treatment consisted of ulcer debridement and offloading of the right foot with a 3D Walker. The patient was seen in November of 2005, 7 months after initial treatment. The right foot still remained dysfunctional with chronic non-healing wounds. Radiographs show radiolucency from mid-shaft of 4th and 5th metatarsal distally to the 4th and 5th distal phalanges and thickened periosteam of the proximal end of the mid shafts of 4th and 5th metatarsals on Figure 13, suggesting osteomyelitis.
Figures 11 and 12 Dorsolateral view of right foot showing ulcer along 5th metatarsal and medial view of the right foot showing cavovarus deformity.

Figure 13 AP Radiograph of bilateral feet.

Figures 14 and 15 Preoperative view of the right foot. Intraoperative picture of the right foot after Lisfranc amputation.

Figure 16 Dorsolateral view of right foot after amputation.

Due to the lack of access to other diagnostic tools, combined with the request from the patient for a permanent solution, a LisFranc amputation was performed on the right foot and osteoset beads with vancomycin were inserted to treat the infection (Figures 14 and 15).

The patient was seen in February of 2006, 3 months after LisFranc amputation of the right foot. The surgical site healed well with good results (Figure 16). However, the cavovarus deformity remained on the left foot (Figure 17).

The patient was seen again in November of 2006 for the last time, 12 months after LisFranc amputation of the right foot. The patient redeveloped an equinus deformity of the right foot and ulcers under styloid processes bilaterally (Figure 18). The patient was treated with well-padded plastazote ankle foot orthosis (AFO).
The Foot and Ankle Online Journal 13 (4): 8

Figure 17 Dorsolateral view of left foot.

Figure 18 Ulcer under styloid process of the right foot after amputation.

Results

The most common lower extremity manifestation was ulcerations. In 17 ulcerated limbs, 8 were insensate and 5 developed osteomyelitis. Out of the 40 limbs, 5 ulcerated limbs had no reported gross deformity, and therefore were not included in the graphs. The remaining 35 limbs were biomechanically classified as rigid and flexible. In the 13 rigid limbs, there were 4 equinus, 3 talipes calcaneus, 7 cavus, and 2 planus feet. These were further subclassified into 6 more categories: equinovarus, pes cavocalcaneus, cavovalgus, cavovarus, pes planovalgus and no additional deformity. Rigid deformities subgroups can be seen in Graph 1. In the 22 flexible limbs, there were 9 equinus, 5 talipes calcaneus, 6 cavus, and 2 planus feet.

Graph 1 Rigid deformity of the foot.

Graph 2 Flexible deformity of the foot.

They were then subclassified into 5 more categories: calcaneovalgus, calcaneovarus, equinovarus, cavovarus, and no additional deformity. Flexible deformities are illustrated in Graph 2.

Discussion

Lower extremity manifestations due to spina bifida are difficult to be classified and the rate of misdiagnosis and mistreatment is high [9]. Similarly to previous findings, the most frequent foot deformity in our study was flexible equinus [4,5,10]. However, we found that rigid pes cavus was the second most predominant foot deformity in the Yucatan Peninsula, contrary to previous reports. Based on the limited medical access in the area and the higher average age of patients presenting with rigid deformities (15.4 years) when compared to flexible deformities (8.8 years), we suggest that this is possibly due to years of leaving the deformity untreated.

The prevalence of spina bifida was 7.85 per 10,000 births or 0.0785% in the country of Mexico [1]. However, in the Yucatan Peninsula the prevalence was found to be significantly higher. In this study, out
of the 1,489 total cases analyzed from years 1999 to 2020, 25 patients with spina bifida were identified. This shows a prevalence rate of 167.90 per 10,000 births or 1.68%. Folic acid is a nutrient that is essential to the development of the fetus. Spina bifida and other birth defects form within the initial 28 days after conception. These congenital deformities can be prevented by ensuring sufficient blood folate levels in the mother during fertile years and early fetal development [11]. In the literature, North America has been shown to have the lowest incidence of spina bifida while Asia has the highest incidence. This could be due to Canada and the United States being the first countries to mandate folic acid fortification. In addition, even though mandatory fortification with folate has been implemented in many countries, it might not be enough folic acid to reach the daily recommended dosage of 400 micrograms. Therefore, it is important for mothers before conception and in the early fetal developmental months to supplement their folic acid intake [12].

Case 1 illustrated a patient with talipes equinovarus, also known as clubfoot deformity on the left foot. This triplanar deformity includes 3 components: ankle equinus, hindfoot varus, and forefoot adduction. Traditionally, there is a higher prevalence of clubfoot in males with a ratio of 2:1 to female and approximately 50% of the cases are bilateral. A few etiologies have been described in the literature, mainly divided into idiopathic and non-idiopathic. Idiopathic consists of limited intrauterine position due to a larger size of the fetus or smaller frame of mothers, while non-idiopathic includes a history of congenital deformities such as spina bifida, cerebral palsy, and meningitis. Clinical presentation of patient 1 at birth predisposed him to a higher risk of developing clubfoot given he is a male with a history of spina bifida. We do not have additional history of patient 1 such as birth weight and height, however, these factors could have also played a role in the patient developing clubfoot [13]. The Ponseti technique, a conservative treatment, was attempted when the patient was 2 years old; however, this technique is only proven to be successful in patients with flexible clubfoot up to 120 days of age [14]. On the right foot, the patient has had a long-standing complex skewfoot with forefoot adduction, midfoot abduction, and calcaneovalgus. The unsuccessful result of the Ponseti method on the left foot, combined with years of the patient not returning for medical assistance, led the bilateral deformity to become rigid on both feet.

Case 2 presented a 2-year-old female with history of spina bifida, insensate feet, active ulcer on the right foot, and flexible bilateral cavovarus deformity. Cavovarus involves a high longitudinal plantar arch, hindfoot varus, forefoot equinus, and pronated first ray in the stance phase of gait. If this deformity is present bilaterally, the most likely etiology is a neurological condition; however, if it is present unilaterally the etiology can be related to trauma such as pilon fractures or talar neck fractures [15]. In a flexible cavovarus foot, surgical correction could be achieved through extensive plantar release and metatarsal osteotomies. However, at the time of the patient’s first visit, physicians from the Yucatan Project prioritized the management of the ulcer prior to correcting any gross deformity. Due to the long period of treatment for ulcer management and limited medical access in the Yucatan Peninsula, the patient did not seek medical help for many years. Recent literature has described that if left untreated, cavovarus deformity can progress into fibrosis of the plantar fascia, shortening and tightening of the achilles tendon leading to excess pressure under metatarsal heads, overloading of the lateral aspect of the foot leading to stress fractures of the 5th metatarsal and more rarely, the cuboid. In addition, it can cause inadequacy of the lateral ligaments and tendons leading to instability of anterolateral ankle and lateral talus [15]. During the 2020 Covid-19 pandemic, we reached out through social media and discovered the patient was no longer ambulating. The patient described a rigid deformity with insensate feet and showed interest in undergoing another surgery, so she could possibly walk again. In a mature foot, surgical intervention might require aggressive techniques including midtarsal osteotomies, calcaneal osteotomies and triple arthrodesis [16]. Final decision for a surgical procedure will only be done in person once full updated history and radiographs are taken.

Case 3 showed the most severe result that could come from insensate lower extremity in spina bifida patients if left untreated for long periods of time: amputation. This patient was first seen at 24 years old, when his rigid cavovarus deformity was present since birth. This deformity caused chronic non-healing wounds that developed into osteomyelitis. Osteomyelitis can be defined as an infectious agent which causes inflammation of the bone. The hallmark of chronic osteomyelitis is the progression of inflammation to tissue necrosis and destruction of bone trabeculae and bone matrix caused by an infectious agent. This is usually accompanied by fragments of bone lacking
blood supply which can become separated to form sequestra and continues to host and spread bacteria despite antibiotic treatment. The fifth metatarsal, first metatarsal, calcaneus, and first digit distal phalanx are the four structures with the highest incidence of developing osteomyelitis in the foot [17]. This case emphasizes the need of spina bifida patients with concomitant lower extremity deformities to seek medical help at a young age to avoid the progression of the deformity and consequently loss of a limb.

Given that this study was built from reports of medical mission trips that occur four times a year with limited resources and time, the lack of documentation of treatment plans and follow-ups made it difficult to identify surgical procedures and assess the success of surgeries. Additionally, we did not have access to the patients birth records or their mothers medical records to accurately determine the etiology of their deformities.

**Conclusion**

The types of foot and ankle deformities seen in spina bifida are diverse in etiology, age and gender of the patients. We discovered the most common lower extremity manifestations of spina bifida in the Yucatan Peninsula are flexible equinus and rigid pes cavus. The mean age of patients with rigid deformities was almost twice as the mean age of the patients with flexible deformities. Zang, et al., concluded that equinovarus requires immediate treatment while valgus deformities can have delayed intervention [15]. Based on our data, we conclude that intervention should be considered as early as possible in any flexible deformity to prevent them from becoming rigid.

**Acknowledgements**

We would like to thank all attendings, residents and students involved in the Yucatan Crippled Children Project along with the International Foot & Ankle Foundation for Education and Research. Additionally, we would like to thank the local Red Cross Hospital in the city of Merida.

**References**