Congenital Talipes Equinovarus: A Case Report Of Bilateral Clubfoot In Three Homozygous Preterm Infants

Abstract

Congenital talipes equinovarus, or clubfoot, is the most common congenital abnormality that over past decades has been diagnosed and treated by various modalities with varying degrees of clinical success. Today it is not only clinicians and physicians who desire to understand the details of diagnosis and treatment, but also the parents of affected children, not only because of the inherent economic and emotional burden of a clubfoot diagnosis but also for proper compliance with treatment protocols to prevent future reoccurrences. This case study outlines the various etiologies, diagnostic modalities, and treatment methods of an incidence of bilateral clubfoot, which presented in three homozygous male preterm triplets.

Introduction

A common congenital deformity of the musculoskeletal system is known as clubfoot, or talipes equinovarus, in which one or both infant feet are excessively planter flexed and swing inward, such that the soles face each other medially (see Figure 1). This deformity can occur in 1 to 3 of every 1 000 Caucasian live births and the severity can range widely among each birth.$^{1-3}$ In the majority of clubfoot cases, infants appear with only the deformation of the foot, or feet, and present clinically without any other grossly visible deformations. This clinical presentation is termed medically as isolated clubfoot, idiopathic clubfoot, or noncomplex clubfoot, with each term being synonymous and interchangeable with the others. The remaining minority of clubfoot cases are termed most commonly as complex clubfoot and present associated with other congenital abnormalities of the newborn including most commonly, but not limited to, spina bifida.$^{1-3}$ Most clubfoot cases are of a male predominance and present bilaterally, but when clubfoot presents unilaterally, most often the right leg and foot are affected as opposed to the left leg and foot. Females, although presenting with a statistically lower birth prevalence for clubfoot, are more likely to pass the deformity on to their children than are males, and females are also more likely to have siblings with the disorder than are affected males.$^2$
Etiology and Pathogenesis

The exact cause of clubfoot is unknown, but current theories suggest the deformity may be caused by genetic (intrinsic) factors or by environmental (extrinsic) factors. Intrinsic factors tend to produce symmetrical bilateral clubfoot and often are associated with additional clinical findings. Such intrinsic factors can arise from many different origins, including genomic additions such as Trisomy 18, genomic deletions such as del(17q23), neurologic malformations such as spina bifida, muscular conditions such as myotonic dystrophy, and connective tissue diseases and disorders including arthrogryposis. Extrinsic factors tend to be most commonly asymmetric in severity although both feet can still be affected to varying degrees. Most commonly extrinsic factors that impede the normal growth and position of the fetus in the maternal womb are implicated in congenital deformities such as clubfoot, and these include specifically uterine constriction due to scarring and fibrosis from multiple prior gestations, a breech position of the fetus in utero, amniotic bands, and oligohydraminos.\textsuperscript{3,4}

Specific Anatomy of Clubfoot

Clubfoot is anatomically characterized and defined via equinus, varus, adduction, and supination of the fetal foot. Equinus is a fixed position of plantar flexion of the ankle joint measured from the attitude of the forefoot in relation to the rearfoot. Varus denotes a deformity in which the angulation of the fetal foot is towards the medial plane of the body. Adduction refers to the movement of the fetal limb and foot towards the midline axis of the body. Supination of the foot specifically refers to a movement or position in which the medial margin of the foot, or the longitudinal arch, is raised. From these characterizations clubfoot is therefore understood in conglomerate to be a subluxation of the talo-calcaneo-navicular joint with underdevelopment of the fascia and soft tissue elements on the medial side of the foot. Frequently, underdevelopment of the calf and peroneal muscles are also seen, which commonly results in shortening of and fibrosis in the gastrocnemius, soleus, and posterior tibial muscles and their respected tendons. The ligamentous structures of the fetal foot are however unaffected in clubfoot deformities, although the bones of the fetal foot are often malpositioned and held in relatively fixed, stable positions by the strong and properly formed ligaments.\textsuperscript{5} This is the anatomical basis for the resistance to clinical realignment.
Prenatal Diagnosis of Clubfoot

For many years conventional radiography was used to assess clubfoot, however it has limited value because the bones in the feet of the child have not yet ossified and are therefore not radiopaque on plain radiographic films. The techniques used in ultrasound are still evolving in the prenatal determination of clubfoot, but ultrasound provides the observation of the cartilaginous components of the tarsal bones which are missed with conventional radiography. Currently, both transvaginal and transabdominal ultrasound are being used for prenatal clubfoot diagnosis in clinical practice (see Figure 2). Transvaginal ultrasound can detect the abnormality for a prenatal diagnosis of clubfoot as early as 12-13 weeks gestation, and transabdominal ultrasound can detect the abnormality as early as 16 weeks gestation. Recently, fetal magnetic resonance imaging (MRI) has been investigated for enhanced detection of clubfoot, but thus far has only seen real utility in patients presenting with complex clubfoot associated with a myelomeningocele. Fetal MRI has been found to be more sensitive than transvaginal and transabdominal ultrasound in the prenatal detection of clubfoot by 18 weeks gestation, but when the diagnosis can be made with ultrasound alone prior to that time point it has been shown that fetal MRI adds no additional information. Improvements in ultrasound therefore have increased the detection of prenatal anomalies such as clubfoot, but no modality has yet been shown to accurately predict the postnatal severity of the clubfoot deformity. The ability to accurately diagnose a clubfoot deformity in utero is important however for the emotional and economic planning of the mother in relation to the necessary postnatal treatment modalities currently employed for treating clubfoot, thus prenatal ultrasound has remained the modality of choice for prenatal diagnosis.

Postnatal Treatment of Clubfoot

Determining Severity

The Pirani scoring system is a standardized clinical system used to assess the initial clubfoot condition and it serves as a guideline in the treatment of clubfoot as it helps to quantify the severity of each clubfoot. Furthermore, it’s objectivity in diagnosing severity and its clinical validity can be reinforced by postpartum ultrasound assessment used before and after each treatment to confirm the efficacy of changes made from each treatment. The Pirani score consists of six anatomical signs, each graded as zero, one-half, or one, with zero indicating no
abnormality and one indicating a severe abnormality. The six anatomical signs are separated further into two sets of three, with one set of three relating to the anatomy of the hindfoot and the other set of three relating to the anatomy of the midfoot (see Figure 3). The higher the Pirani score, the more severe the clubfoot deformity, yet the Pirani score is not a static clinical assessment technique; instead the affected foot or feet should be constantly rescored using the Pirani method both before each casting change or physical manipulation treatment as well as after. An ideal treatment modality would see a gradual yet predictable lowering of the Pirani score until a score of zero was reached.

*The Ponseti Method*

Regardless of the severity of the deformity, there are different methods of treatments available. Methods that are currently used include serial casting and manipulation treatments, but the outcomes of each are enhanced if they are used as early as possible, and in some cases of clubfoot, proper treatment will still require surgical correction regardless of the method chosen. Surgical correction of clubfoot was initially thought to offer superior results instead of the then most clinically used method of casting and manipulation, called the Ponseti method of manipulation (see Figure 4). “Many complications were encountered with operative treatment such as forefoot adduction, cavus, overcorrection, hindfoot equinus or varus, and avascular necrosis of the talus.” The Ponseti method was not accepted as a viable surgical alternative at first due to its high reoccurrence rate, but was revived later after the complications of operative treatments.

The Ponseti method stresses the use of postcasting abduction splints, also known as foot abduction orthoses, and physical manipulation of the foot via splinting to achieve and maintain correction, which is crucial in preventing a reoccurrence of the clubfoot after the removal of the last cast. To begin the Ponseti method, the medial ligaments of the foot are stretched and held in place by casts. This process is then repeated as necessary with a series of casting and recasting, until the foot reforms into proper anatomic alignment, typically within four to six weeks although it can take longer for cases of clubfoot to achieve proper correctional alignment. After the last cast is placed, a percutaneous Achilles tenotomy, a simple surgical procedure to cut a portion of the Achilles tendon to relieve muscular tension on the talus caused from the underdeveloped length of the posterior leg musculature, is used to correct any residual equinus.
and to acquire a dorsiflexion of 15-25 degrees, which is the required standard for current treatment.\textsuperscript{7}

\textit{French Taping Method}

The French taping method (see \textbf{Figure 5}), along with the Ponseti method of manipulation, is currently becoming the most widely practiced treatment of clubfoot. The French taping method uses specially trained physiotherapists to do almost daily manipulations and structural bracing with taping, requiring one hour physical therapy sessions five days a week. This is continued every day and every week until the infant begins walking, at which point the manipulations and taping are discontinued as the taping immobilizes the muscles around the feet and would therefore cause fetal injuries in infants learning to walk. The French taping method does not eliminate the need for surgery or Achilles tenotomy procedures however, which is why it is most often used as an adjunct to, and not a replacement of, the Ponseti method.\textsuperscript{8}

\textit{Ilizarov Technique}

For severe foot deformities with very high Pirani scores greater than five, for affected feet that have been neglected for early-onset treatment, and for recurrent clubfoot deformities, the Ilizarov technique is used as an alternate to the conventional Ponseti method and French taping technique (see \textbf{Figure 6}). The Ilizarov technique reduces the risks of not only shortening of the foot but also of neurovascular and cutaneous complications, yet it requires invasive surgery. This method approaches treatment in a two-step application, one directed to treatment of the foot and the other directed to treatment of the leg. This technique realigns the proper joint surfaces with the proper foot anatomy by placing tension wires through the boney structures of the leg and foot. After the wires are removed a plaster cast is worn followed by an ankle-foot orthosis (see \textbf{Figure 7}).\textsuperscript{9} Due to the invasiveness of the surgeries required for wire placements and surgical complications in newborn infants, the Ilizarov technique is not the current standard of care for cases of clubfoot with low Pirani scores but is reserved for only severe, neglected, or recurrent cases.

\textit{Postcasting Foot Abduction Orthoses}

After manipulation and casting of the Ponseti method, it is important to use postcasting abduction splints or braces known as foot abduction orthoses. There are several different types of foot abduction orthoses that can be used in the treatment of clubfoot and they all have several
features in common. They all consist of some form and shape of a bar with a shoe of some type attached approximately shoulder width apart that holds the affected foot, or feet, in 70 degrees of external rotation, while an unaffected foot if present is held in 40 degrees of abduction. These new foot abduction orthoses and their varying designs are improving success rates of treatment and reducing reoccurrence rates of clubfoot due to increased compliance of treatment amongst the parents.

One of these new brace designs is known as the Mitchell Brace or Mitchell-Ponseti Brace. This brace includes the standard bar common to all foot abduction orthoses, but with removable shoes with silicon ankle-foot-orthosis type inserts (see Figure 8). This Mitchell-Ponseti Brace was designed to be more comfortable and easier to use with patients who had limited dorsiflexion and with feet that were difficult to brace. The shoe that is used is a developed sandal that has three straps and a rubber like sole that can be put on and worn before the shoe must be inserted onto the bar, making assembly easier on the parent. It contains a window to ensure proper foot placement of the heel for the parents as well. This Mitchell-Ponseti Brace is now replacing the former Dennis Browne Abduction Splint and Markell Brace.

The Dennis Browne Abduction Splint has the shoes directly attached to and irremovable from an L-shaped metal footplate that allows for foot eversion without the calcaneus being ab ducted (see Figure 9). The Markell Brace is another alternative foot abduction orthosis which uses an open toed shoe that is attached to an aluminum spreader bar that is available in many different sizes, which allows the child to maintain compliance with the Markell Brace as it grows (see Figure 10). This brace was recently modified to allow the shoes to be put on without the bar; however, it is very difficult to know if the child’s heel has been placed in the shoe properly. To counter this, there is now a line built into the shoe that allows parents to ensure proper placement, because a simple visualization of the infant’s toes passing the line is enough to indicate that the shoe is improperly placed. Should complications with the line arise, an alternate shoe with a cut out heal can also be used.

The Dobbs Dynamic Brace is more expensive compared to the Mitchell-Ponseti Brace, Dennis-Browne Abduction Splint, and the Markell Brace, but it has some added benefits for treatment and compliance (see Figure 11). This brace allows each leg to have active extension and flexion while keeping the external rotation of the foot that is necessary, allowing proper bracing while also providing some lower limb movement. The shoe is detachable from the bar
via a Velcro strap and it also has custom molded inserts that fit inside allowing for increased ease of assembly and patient customization.

Improper use of any postcasting foot abduction orthoses can lead to skin problems such as skin irritation and ulceration. If the problem is superficial it can be fixed by a modification to the brace such as cutting out a window over the troubled area, interchanging shoe or insert designs if available, or even changing the brace itself. If the problem is due to a full thickness sore or ulceration than discontinuation of the brace must occur, although full thickness sores and ulcerations are uncommon. The brace is then reused again once the sore or ulceration is gone. In the time between castings the feet will often begin to relapse and the sore areas may need treatment or protective covering depending on the cleanliness of the wound.

*Clubfoot Reoccurrence*

The major issue with the Ponseti method early on was its high reoccurrence rate of clubfoot, although since the advent of newer braces and increased treatment compliance showing greater benefit with the Ponseti method, it is quite possible that those early high reoccurrence rates correlated with the lack of compliance, or lack of understanding from the parents regarding their infant’s treatment instead of an inherent flaw in the ability of the Ponseti method to correct clubfoot. Other issues that could contribute to the early high reoccurrence rates are incorrect casting technique, poorly fitting splints, and improper tenotomy. Often parents have a hard time distinguishing a cry of pain from a cry of annoyance and the parents remove the brace. Frequent removal of the brace creates relapses because correct placement of the fetal bones of the foot is hindered, and subsequent castings then cause the infant pain or discomfort, which again leads parents to prematurely remove the brace due to the cries of their child, creating a cycle of noncompliance and relapse of treatment.

To catch the early identification of relapse in clubfoot, it is important for regular follow-up visits after each casting. Once it has been demonstrated to the physician or clinician that the brace is being used properly and with correct patient compliance, the patient is seen at that point again only after six weeks, and then again after 12 weeks of full-time use. After the allotted time for full-time use of the brace the infant is switched to naptime and nighttime use and they are followed up with every three to four months. This can be extended to a follow up every four to six months depending on the reliability of the parents with treatment compliance.
Case Study

A 35-year-old woman was referred at 33 weeks gestation to a tertiary care unit for a threatened preterm delivery. At 34 weeks gestation she gave birth to male triplets by cesarean section, which was elected due to a history of multiple pregnancies, having had a previous cesarean section, and current uterine contractions. Her history was negative for any associated risk factors of oligohydramnios or smoking. During her first trimester, an ultrasound examination identified the woman was pregnant with identical triplets in a triamniotic monochorionic pregnancy, and succeeding ultrasound examinations showed both symmetrical as well as normal growth rates of the fetuses, with no presence of structural anomalies detected.

At the time of birth, the Apgar score for all three newborns was 8 at one minute and 9 at five minutes, yet all three infants presented with fixed bilateral talipes deformities despite the prenatal ultrasound examinations detecting none. Immediately following delivery the Ponseti method was begun for treatment, along with manipulation and sequential cast applications. Casting changes were done every week, with the application of gradual abduction-external rotation used to help restore the normal alignment of the midfoot and forefoot. A tenotomy of the Achilles tendon was performed on all three infants when they were three months old, and at 10 months of age a Dennis Browne Abduction Splint was used to maintain proper foot positioning until the infants could walk. For the next two years, the clinicians recommended nighttime splinting to prevent any reoccurrence of the clubfoot.12

Conclusion

The woman in this case study gave birth to three male triplets, all of whom had fixed bilateral clubfoot deformities indicating that all three either shared a common genetic or genomic abnormality or were all subjected to the same extrinsic factor, which could easily be postulated as decreased uterine room for adequate fetal limb development of the triplets. This case study also illustrates a great point regarding the prenatal diagnosis of clubfoot with ultrasound. As discussed above, transvaginal ultrasound has been shown to detect clubfoot as early as 12-13 weeks gestation, and transabdominal ultrasound has been shown to detect clubfoot as early as 16 weeks gestation. This woman received an ultrasound in her first trimester, which with a standard 40 week gestational timeframe places her ultrasound sometime before the 13th week of
gestation. This would have been too early to detect clubfoot with either method. However, it is reported that she received subsequent ultrasound imaging and that no deformities were found, illustrating well the need to develop more sensitive prenatal diagnostic tools for clubfoot.

The Apgar scores for the children were within normal ranges indicating healthy infants at birth. This score is a standardized rating system, similar in function to the Pirani score as a standardized rating system, for fetal health immediately following birth. Multiple areas of the infant are graded on a scale of one to three, and assessments are always made at exactly one minute of life and five minutes of life. A healthy infant should score between a seven and a ten, and the score should not regress between the one-minute measurement and the five-minute measurement. In this case, all three infants were born with healthy Apgar scores. Then, all the infants were subjected to the Ponseti method of treatment, followed by an Achilles tendon tenotomy and finally use of a Dennis Browne Abduction Splint with a physician or clinician recommendation of nighttime splinting to prevent reoccurrence. There was no indication in the case report as to the long-term outcome of these three male infants, but the key to reducing the reoccurrence of clubfoot and achieving a lifelong correction of this deformity lies in proper parental education regarding the correct long-term use of postcasting foot abduction orthoses.
References


Figures and Captions

Figure 2. The plantar surface of both fetal feet can be visualized in this ultrasound, which shows the tibias and fibulas in the same plane as the plantar surface, indicating a diagnosis of clubfoot. Transvaginal and transabdominal ultrasounds produce such images in the prenatal diagnosis of clubfoot. Image courtesy of a local hospital.
Figure 3. The Pirani scoring system is a standardized rating system to quantify the severity of postpartum clubfoot. It is based on a simple scaling system, composed of three variables in the hindfoot and three variables in the midfoot, each of which receives a score of zero, one-half, or one point based upon severity. The higher the Pirani score, the worse the severity of the clubfoot. Image courtesy of Maranho DA, Volpon JB. Congenital Clubfoot. Acta Ortop Bras. 2011;19(3):163-9. Available at http://www.scielo.br/aob. Accessibility verified November 07, 2013.
Figure 4. The Ponseti method of manipulation and casting consists of a series of foot and leg castings for correction of clubfoot, which typically takes place over a period of four to six weeks although longer intervals are not uncommon. Image courtesy of The Clubfoot Club. How is clubfoot corrected? Available at http://clubfootclub.org/about/ponseti-checklist/. Accessibility verified November 04, 2013.
Figure 5. The French taping method utilizes daily physiotherapy and casting along with taping to reform the foot of an infant diagnosed with clubfoot. Today, the French taping method is most commonly used as an adjunct to the Ponseti method of manipulation and serial casting. Image courtesy of The Clubfoot Club. How is clubfoot corrected? Available at: http://clubfootclub.org/about/treatment-options/. Accessibility verified November 04, 2013.
Figure 6. *The Ilizarov technique requires surgical intervention in which wires are placed in the bones of the foot and leg. Tension is then applied to the wires to achieve proper foot reformation of clubfoot. This technique is reserved only for severe, neglected, or recurrent cases of clubfoot due to the need for invasive surgery on the newborn. Image courtesy of The Clubfoot Club. How is clubfoot corrected? Available at: http://clubfootclub.org/about/treatment-options/.* Accessibility verified November 04, 2013.
Figure 7. Ankle foot orthoses are hard plastic casings designed to be worn following the Ilizarov Technique of correction for clubfoot. Image courtesy of The Clubfoot Club. How is clubfoot corrected? Available at: http://clubfootclub.org/about/treatment-options/. Accessibility verified November 04, 2013.
Figure 8. A Mitchell-Ponseti brace for correction of clubfoot, shown with an adjustable bar and removable sandal-like shoe with silicon ankle-foot-orthosis type inserts, designed to be worn post-treatment of clubfoot. Image courtesy of The Clubfoot Club. How is clubfoot corrected? Available at: http://clubfootclub.org/about/treatment-options/. Accessibility verified November 04, 2013.
Figure 9. Dennis Browne Abduction Splints have the shoes directly attached to and are irremovable from an L-shaped metal footplate that allows foot eversion without the calcaneus being abducted. This is an example of a postcasting foot abduction orthosis used in the post-treatment stage of clubfoot to prevent reoccurrence. Image courtesy of The Clubfoot Club. How is clubfoot corrected? Available at: http://clubfootclub.org/about/treatment-options/. Accessibility verified November 04, 2013.
**Figure 10.** The Markell brace uses an open toed shoe that is attached to an aluminum spreader bar that is available in many different sizes, which allows the child to maintain use as it grows. This is yet another example of a post-treatment foot abduction orthosis for the prevention of reoccurrence of clubfoot. Image courtesy of Zionts LE, Dietz FR. Bracing following correction of idiopathic clubfoot using the Ponseti method. *J Am Acad Orthop Surg.* 2010;18(8):486-93.
Figure 11. The Dobbs Dynamic Brace allows each leg to have active extension and flexion while keeping external rotation of the foot for prevention of reoccurrence of clubfoot. Although more expensive than other ankle foot orthoses, it is the only one to maintain a degree of motion in the child. Image courtesy of Zionts LE, Dietz FR. Bracing following correction of idiopathic clubfoot using the Ponseti method. J Am Acad Orthop Surg. 2010;18(8):486-93.