ISSN: 2687-816X
DOI: 10.33552/GJOR.2022.04.000577
Global Journal of Orthopedics Research
Iris Publishers

Case Report

Fibular Hemimelia: A Case Series of Nine Patients Treated with Syme Amputation

Julio Avila[1]*, Jessica A Nelson[1], Selina Silva[2]

[1]*University of New Mexico School of Medicine*

[2]*Univerisity of New Mexico Department of Orthopaedics and Rehabilitation*

***Corresponding author:** Jessica Nelson, University of New Mexico School of Medicine, Albuquerque, New Mexico, USA.

Received Date: September 12, 2022

Published Date: October 12, 2022

Abstract

Fibular hemimelia (FH) is a rare congenital long bone deficiency that can occur with other lower limb deformities. It is important to report on patients with FH because the condition is rare, and there are different recommendations relating to operative treatment. The purpose of this paper is to describe a case series of nine patients with FH, discuss treatment, functional outcomes, rehabilitation potentials, and prostheses. Eight of nine patients in this case series had severe FH, or complete absence of the fibula. All patients underwent a Syme amputation. The patients had good clinical outcomes and potential for rehabilitation based on activity levels. The average number of surgeries 1.8, which includes the Syme amputation. Complications observed include genu valgum and knee laxity. The results of this case series will assist in decision making for pediatric orthopaedic surgeons who have patients with severe FH. When compared to limb salvage procedures, patients with amputations have less surgeries and complications, which is something that should be considered when deciding treatment.

Keywords: Fibular hemimelia; Syme amputation; Lower limb deformities; Longitudinal fibular deficiency; Lower limb prostheses

Introduction

Fibular hemimelia (FH) is the most common congenital long bone deficiency, occurring in between 1 to 135,000 and 1 to 50,000 births [1–3]. Most cases are sporadic, and patients can present with varying degrees of fibular deficiency, ranging from partial absence to complete absence. Regarding the limb distal to the fibula, patients may have concomitant ankle and foot deformities, such as absent lateral rays, tarsal coalitions, calcaneovalgus or varus [1,2]. Proximal to the fibula, individuals may have cruciate ligament deficiencies, knee valgus deformity, or femoral hypoplasia [1,2].

Depending if the cruciate ligaments are hypoplastic or aplastic, the severity of knee instability varies, but typically is asymptomatic in childhood [3]. Multiple classification systems of FH exist. Achterman and Kalamachi is the most common classification system of FH-type Ia- fibular hypoplasia; type Ib- partial absence; type II- complete absence [3]. Paley et al. reports the best prognostic factor of FH is the foot deformity, and the degree of foot deformity does not correlate with the degree of fibular deficiency [1]. The Paley classification system grades deformities of the fibula in conjunction with problems with the ankle and subtalar joint, and advocates for limb lengthening procedures [1]. Birch et al. classified FH based on number of rays and recommends amputation for less than three, versus limb salvage for at least three rays [4]. Our group's experience has been that a fair number of these patients have three rays and there are not clear recommendations in the literature of how to evaluate this group. The two methods to correct FH are limb salvage or amputation. The literature is inconclusive on whether

amputation versus limb lengthening is more favorable [5,6]. The number of rays a patient has may lead the surgeon to choose one treatment option over the other.

This case series presents nine patients who had a Syme amputation. The purpose of this case series is to increase existing literature on FH treatment. Patients with FH at our institution who have been treated with Syme amputation have satisfactory clinical outcomes and acceptable rehabilitation potentials, or activity levels.

Method

This is a retrospective case series that received institutional review board approval. Clinical records, radiographs, functional data, and prosthesis type were reviewed. The Achterman and Kalamchi classification system, radiographs, and clinical exam for FH was used to determine severity of FH for the patients in this case series. Severity was assessed via presence of the fibula, partial absence of the fibula, complete absence of the fibula, and whether the distal fibula is present and supports the ankle.

Inclusion Criteria: Patients treated at the University of New Mexico Carrie Tingley Hospital from 2006 to 2018 with a diagnosis of FH were included. They had to be less than 18 years old at the time of presentation. We did not exclude any patient that was an adult by final follow-up. We selected as far back as 2006 because that is when we started collecting digital imaging and required at least a two-year follow-up. Patients did not need a Syme amputation to meet inclusion criteria, only a diagnosis of FH, it just so happens that all the patients in this series since 2006 under the age of 18 have been treated with Syme amputation.

Exclusion Criteria: We excluded patients that do not have a follow-up of at least 2 years or patients who underwent a Syme amputation for other reasons. We did not exclude patients who underwent salvage and subsequent limb-lengthening procedures. There were no patients at our institution who had limb salvage procedures.

Case Series Presentation

Eight patients out of nine had severe Type II FH on at least one limb. One patient had a milder case of FH, classified as Type 1a. Two patients had bilateral FH. Table 1 describes patient characteristics and Table 2 shows details of prostheses. One patient had FH in conjunction with VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anomalies, and limb abnormalities) and another had FH in conjunction with femoral hypoplasia, and unusual facies syndrome (starred in Table 1 and 2). Syme amputation was performed as described by Hudson et al and is done when the patient is approximately one year old. Figure 1 shows a patient with 2 rays who had complete absence of the fibula, and thus the original surgeon believed amputation was appropriate. This speaks to the topic of discussion mentioned in the introduction of how the number of rays can influence the decision to amputate. These patients and their families meet with their prostheses team and Orthopaedic surgeon several times before their surgery to improve patient comfort with informed decision making (Table 1&2).

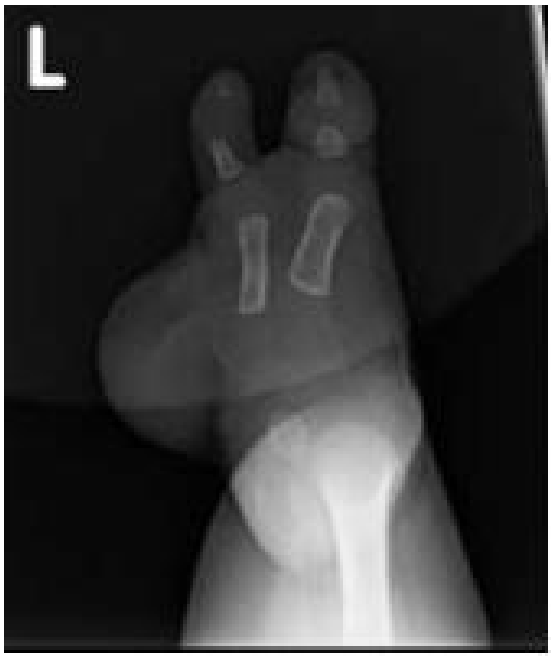

Figure 1: Dorsoplantar view radiograph showing a patient in the case series with two rays and an absent fibula.

Table 1: Description of Case Series.

S.No	Type of FH (Achterman and Kalamachi Classification System)	Sex	Laterality	Concomitant Lower Limb Deformities	Age at Syme Amputation + other procedures	Follow-up Procedures	Latest Follow-Up	Current Age
1	Type IA	M	R	Four rays, clubfoot deformity (with varus hindfoot, equinus and supination of his forefoot), tibial shortening, tarsal coalition in subtalar joint	17 months, Syme + Achilles tendon lengthening (2006)	2017- b/l medial distal femur hemi epiphysiodesis, 2019- Removal of deep femur implants	2020- prosthesis doing well, b/l genu valgum, right knee reproducible "popping" in the anterior lateral region (negative anterior drawer test, stable to valgus/varus stress, general increased laxity in knee)	17
2	Type II	M	R	four rays, shortening of right tibia, tarsal coalition	13 months (2007)	None	b/l genu valgum, lost to follow-up 2014, doing well with prosthesis	15
3	Type II	F	L	three rays	9 months (2007)	None	2010- concerns for valgus growth at distal aspect of residual limb, lost to follow-up thereafter, doing well with prosthesis	15
4	Type II	M	L	knee laxity, fixed valgus and equinus of ankle, rigid empty hindfoot, syndactyly of great and second toe	12 months (2007)	None	2007- lost to follow-up	15
5	L- Type II, R- Type IA	F	B/L	L- two rays, fused hindfoot; R- three rays, stable plantigrade foot and ankle with palpable lateral malleolus	12 months left Syme (2010)	2020- left distal tibia drill epiphysiodesis; 2021- left proximal tibial epiphysiodesis	2021- difficulty fitting prosthesis, 2 weeks post-op most recent procedure -mild left genu valgum	12
6	Type II	F	L	three rays, calcaneal valgus deformity	15 months (2015)	None	2021- needing a new prosthesis otherwise doing well, mild genu valgum b/l	8
7	L- Type II, R- Type IA	M	B/L	L- two rays, severe equinus and valgus deformity of ankle joint with a bend in the tibia; R-four rays, plantigrade flexible, full ROM at knee and ankle	13 months (2016)	2021- surgical removal of bony prominence of anterior left tibia that caused pain, Achilles tenotomy because heel pad appears to be shifting backward	Surgery in 2021 referred to in 'follow-up procedures -mild left genu valgum 2020	6
8*	Type II	F	R	R proximal focal femoral deficiency with coxa vara, left dysplastic and subluxated hip, left equinus contracture	17 months (2006)	2008- left open hip reduction with capsulorrhaphy & right contralateral valgus producing osteotomy 2009- removal of implant right hip 2011- right coxa vara recurred, valgus producing osteotomy of the femur with derotation 2013- Left Achilles tendon lengthening	2021- due to bilateral congenital hip dysplasia, patient considering undergoing arthroplasty. -Prosthesis doing well.	16

9*	Type II	M	L	L Complete FH with proximal femoral focal deficiency, L varus foot deformity, teratogenic hip dislocation	27 months (2011)	-2015- Arthrodesis: of knee with plate screws and resection of distal femur and proximal tibia -2015 repeat arthrodesis of knee with intramedullary nail and neutralizing antirotation plate (to better tolerate prosthesis)	2021- Prosthesis doing well. The patient has severe scoliosis so gets around with assistance of forearm crutches	12

*Patient 8: Femoral Hypoplasia and Unusual Facies Syndrome
*Patient 9: VACTERL Syndrome

Table 2: Number of New Prostheses, Number of Broken Feet, and K-levels.

Patients	Year of Syme & Age	Type of Prosthesis & Total # New Ones	Broken Feet	Current Age	K-level
Patient 1	2006 at 17 mo.	Syme, 7 total • 2006- First • 2013- New • 2014- New • 2016- New • 2018- New • 2020- New • 2021- New (Received care outside of hospital between 2006-2012)	Multiple Broken Feet (3)	17	K3
Patient 2	2007 at 13 mo.	Syme, 7 total • 2007, First • 2017, New • 2010, New • 2011, New • 2012, New • 2013, New • 2013, New (No follow-up beyond 2014)	Multiple Broken Feet (2)	15	K3
Patient 3	2007 at 9 mo.	Syme, 3 total • 2007, First • 2007, New • 2009- New (No follow-up beyond 2009)	None	15	K3
Patient 4	2007 at 12 mo.	Syme, 1 total • 2007, First No follow-up beyond 2007	None	15	K3

Patient 5	2010 at 12 mo.	Syme, 8 total • 2010, First • 2011, New • 2012, New • 2014, New • 2016, New • 2018, New • 2019, New • 2020, New	Multiple broken feet (5)	12	K4
Patient 6	2015 at 15 mo.	Syme, 6 total • 2015, First • 2015, New • 2017, New • 2018, New • 2020, New • 2021, New	None	8	K4
Patient 7	2016 at 13 mo.	Syme, 1 total • 2016, IPOP • 2016, Syme	None	6	K3
Patient 8* (FHUFS)	2006 at 17 mo.	Syme • 2006, First • 2007- New • 2008, New • 2010, New • 2011, New • 2012, New • 2014, New • 2017, New • 2021, New	None	16	K2
Patient 9* (VACTERL)	2011 at 27 mo.	Above Knee (AK) Prosthesis • 2011, First • 2016, New • 2018, New • 2019, New • 2021, New	None	12	K3

At last follow-up, all patients in this case series were ambulatory with acceptable rehabilitation potential, as defined by K-Levels. K-levels refers to a rating system that is used by Medicare to indicate a person's potential functional ability who has a prothesis. Most of the patients performed age-appropriate activities that included walking, running, and jumping and had an average of a K3 level. Acceptable limb alignment was determined due to the lack of patient complaints of limb alignment signs like painful or unbalanced walking, at last follow-up. Patients 8 and 9, who had a syndromic condition, had lower activity levels. Five of nine patients had unilateral or bilateral genu valgum as a complication of FH. One patient received a corrective hemiepiphysiodesis for their genu valgum, two other patients are planned to have a hemiepiphysiodesis around age ten if their genu valgum does not correct, and the fifth patient is utilizing physical therapy and prosthesis modification for slight genu valgum.

Common reasons for protheses follow-up after Syme amputation is for comfort issues, broken prosthetic foot, or replacement. Eight of nine patients used a below knee Syme hard socket stovepipe design prosthesis. Patient nine required an above knee prosthesis in the setting of knee arthrodesis and proximal femoral focal deficiency. This patient's arthrodesis was performed to correct the misalignment and severe flexion of their remaining

limb to increase limb functionality. The foot attached to the distal end of the prosthesis depends on the activity of the patient. More active patients received prosthetic feet that were less stable (not fixed in a position) but had axes of rotation for ankle inversion and eversion.

In this case series, patients 1, 2, 5, 6, 7, 8, and 9 have at least 5 years of follow-up. For these seven patients at 5 years from their Syme amputation, the average number of procedures per person (including amputation) was 1.8 with a range of 1 to 4. Patients 3 and 4 were lost to follow-up at approximately 3 years and 1 year from their Syme amputation, and at those time frames, did not require additional procedures.

Every patient frequently had their prosthesis to match their growth and to try to avoid genu valgum of the knee. Most patients only need a new prosthesis every year or two because of prosthetists altering their external hardware for growth accommodation. A prosthesis that is too short was a common complaint. It may be more cost effective to alter the prosthesis as much as possible before ordering new pieces after growth surpasses the ability of the prosthetics lab to ensure functional accommodation. Multiple patients had foot breaks and replacement prosthetic feet.

Discussion

This case series indicates that patients undergoing Syme amputation have good clinical outcomes and acceptable rehabilitation potential based on activity levels. There is a lack of studies describing functional potential of patients undergoing amputation versus salvage surgery, so this case series is important to highlight success after amputation. A consistent way to report functional outcomes remains a challenge with this patient population as K-levels have not been strongly proven to connect to patient outcomes, only to their rehabilitation potential. A limitation of this case series is not including a cohort of patients who underwent limb salvage to compare outcomes. However, there were no patients at our institution who had a limb salvage procedure, so this was not possible.

Naudie et al. reported that amputation for severe FH is more favorable with less complications when compared to limb lengthening procedures [5]. A study by McCarthy et al. found that amputation patients had an average of 1.9 procedures compared to 7 procedures in the lengthening group at an average follow-up of 7 years [6]. The average of 1.8 procedures reported in this case series is consistent with the current literature.

Some authors like Paley et al. advocate for limb reconstruction.1 Multiple studies have described favorable outcomes with limb sparing reconstructive surgery [7,8]. In less severe forms of FH, Changulani et al. concluded that limb reconstruction is a good option with satisfactory clinical outcomes [9]. Most surgeons agree that in cases of FH with mild to moderate leg length discrepancy (LLD) with a mild to moderate foot deformity that these patients have better outcomes with reconstruction and limb lengthening procedures[3]. With severe LLD and severe foot deformity, amputation is preferred because of the failure to obtain acceptable outcomes after limb lengthening.3

Genu valgum and knee laxity due to likely cruciate deficiency were complications observed clinically. These patients reported their symptoms as mild, but physical exam did show laxity. No patients underwent cruciate ligament reconstruction. Figure 2 shows an anteroposterior radiograph of a patient who had a bilateral medial distal femur hemiepiphysiodesis 11 years after Syme amputation to correct genu valgum Figure 2. Further research is needed in this population to know if knee ligamentous reconstruction would be of benefit (Figure 1&2).

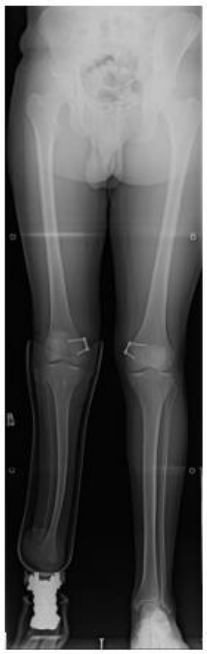

Figure 2: Anteroposterior radiograph of a patient in the case series who had a bilateral medial distal femur hemiepiphysiodesis 11 years after Syme to correct genu valgum.

In cases of severe FH, as seen in this study, patients with Syme amputation do well with minimal complications and maintain acceptable levels for potential rehabilitation. Genu valgum is one of the most common complications and was seen in five patients. Several patients also had anterior-posterior knee laxity on clinical exam, but they have not required ligamentous reconstruction. Satisfactory clinical outcomes in this case series are indicated by patients having an average of less than 2 associated surgeries including the Syme amputation and acceptable potential for rehabilitation defined by having a K2 or greater level. Six of nine patients still follow-up at our hospital to date and the other three are receiving care at a different facility. Two patients had FH in conjunction with a syndromic condition, which makes interpreting their outcomes difficult, as poor outcomes could be due to multiple factors related to their diagnosis. However, it is important to include these two patients so orthopaedic surgeons are aware of FH occurring in conjunction with other non-orthopaedic pathology.

Acknowledgement

A special thank you to the University of New Mexico Carrie Tingley Orthotics and Prostheses Team.

Conflict of Interest

The authors report no conflict of interest.

References

1. Paley D Surgical reconstruction for fibular hemimelia. J Child Orthop 2016;10(6):557-583.
2. Morris EJ, Tofts L, Patterson M, et al. (2020) Physical performance of children with longitudinal fibular deficiency (fibular hemimelia). Disabil Rehabil. Published online December 17:1-11.
3. Fuller CB, Shannon CE, Paley D (2021) Lengthening Reconstruction Surgery for Fibular Hemimelia: A Review. Children (Basel):8(6):467.
4. Birch JG, Lincoln TL, Mack PW, Birch CM (2011) Congenital fibular deficiency: a review of thirty years' experience at one institution and a proposed classification system based on clinical deformity. J Bone Joint Surg Am 93(12): 1144-1151.
5. Naudie D, Hamdy RC, Fassier F, Morin B, Duhaime M (1997) Management of fibular hemimelia: amputation or limb lengthening. J Bone Joint Surg Br 79(1): 58-65.
6. McCarthy JJ, Glancy GL, Chnag FM, Eilert RE (2000) Fibular hemimelia: comparison of outcome measurments after amputation and lengthening. J Bone Joint Surg Am 82(12):1732-1735.
7. El-Sayed MM, Correll J, Pohlig K, (2010) Limb sparing reconstructive surgery and Ilizarov lengthening in fibular hemimelia of Achterman-Kalamchi type II patients. J Pediatr Orthop B 19(1): 55-60.
8. Mishima K, Kitoh H, Iwata K, et al. (2016) Clinical Results and Complications of Lower Limb Lengthening for Fibular Hemimelia. Medicine (Baltimore) 95(21): e3787.
9. Changulani M, Ali F, Mulgrew E, Day JB, Zenios M (2010) Outcome of limb lengthening in fibular hemimelia and a functional foot. J Child Orthop 4(6): 519-524.