Prosthesis for a Patient with Proximal Femoral Focal Deficiency: A Case Report

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ABSTRACT

Proximal femoral focal deficiency is a rare birth defect that affects the hip bone and the proximal femur. The incidence is one case per 50,000 to 200,000 population. The disorder may be unilateral or bilateral, with the hip being deformed and the leg shortened. The goal of treatment is to provide optimal function during standing and ambulation. A 15-year-old male diagnosed with left proximal femoral focal deficiency was admitted for prosthetic rehabilitation. He presented with a very short left lower extremity, 38 cm leg length discrepancy, flail left hip and knee joints, and normal range of motion at the left ankle, and with muscles graded at 4/5. The patient was independent in transfer activities and ambulated with bilateral axillary crutches. A combination of orthosis and prosthesis (henceforth "prosthosis") was designed for the patient with a mechanical hinge joint to equalize the leg length and to improve lower extremity function during standing and ambulation. Upon discharge, the patient was independent in donning and doffing the prosthesis, was ambulatory using the prosthesis without gait aid but with minimal listing during the stance phase on the prosthesis side. During the patient's two-year follow-up, adjustment of the prosthesis was done to accommodate growth; checking of the prosthesis for mechanical breakdown and anticipatory management of potential musculoskeletal complications and psychosocial concerns on the use of the prosthesis were also done.

Key Words: Proximal femoral focal deficiency, leg length discrepancy, prosthesis, "prosthosis"

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Introduction

Proximal femoral focal deficiency (PFFD) is a rare birth defect affecting the hip bone and the proximal femur with an incidence of one case per 50,000-200,000 population. It may affect one or both sides, with the hip being deformed and the leg shortened. The etiology is unknown, but theories have been proposed and agents implicated. The sclerotome subtraction theory states that injury to the neural crest cells that form the precursors to the peripheral sensory nerves of L4 and L5 results in PFFD.[1,2] Boden et al. reported that PFFD results from a defect in the proliferation and maturation of chondrocytes in the proximal growth plate.[3] Possible causes include anoxia; ischemia; irradiation; bacterial and viral infections; toxins; hormones; and mechanical, chemical and thermal injury.[2] Thalidomide, when taken between the fourth and sixth weeks of gestation, has been demonstrated to be a definite cause of PFFD in humans.[4] Currently, no evidence indicates a genetic etiology.[1]

The goal of treatment in PFFD is to provide optimal function during standing and ambulation. Both non-surgical and surgical treatments are available. The use of prosthesis is a relatively inexpensive and effective solution but there are limited outcome studies on prosthesis use. The design presented in this case report is a modification of the hybrid prosthesis with a mechanical hinge joint that acts as a knee to allow it to flex during sitting. A manual locking mechanism keeps the knee extended during ambulation. The prosthesis equalizes the leg length and increases control of the proximal muscles acting on the hip to improve lower extremity function during standing and ambulation in individuals with PFFD.

Case Report

A 15-year-old male from Laguna was admitted to a tertiary government hospital in May 2008 with a chief complaint of significant leg length discrepancy. He was born full term to a 24-year-old G2P1 (1001) mother via normal spontaneous vaginal delivery in a hospital with no fetomaternal complications noted. At birth, shortening of the left lower extremity was noted. The mother had regular prenatal care and denied exposure to radiation and chemical toxins. However, she had a urinary tract infection in the ninth month of pregnancy and was given unrecalled

medications. Developmental milestones were at par with age except in ambulation. Parents refused surgical correction of the lower extremity deformity. At four years of age, the patient was prescribed an extension prosthesis that enabled him to stand and ambulate independently (Figure 1). Length adjustment of the prosthesis was done every six months to accommodate the height of the patient until he was eight years old. Since then, the patient ambulated without prosthesis but with bilateral axillary crutches. At age 14, he was brought to an orthopedic surgeon for surgical correction. Van Ness rotationplasty and Syme's amputation were offered but surgery was deferred until he reached full skeletal maturity. The patient was then referred to Rehabilitation Medicine for intervention and was admitted to the Rehabilitation Medicine ward.

Past medical history and family history were unremarkable. He is the second of five children, and a third year high school student in a private school with average academic performance. He lives with his mother, a housewife, and four siblings in a two-storey house. His father is employed. His bedroom is located on the first floor approximately 7 meters away from the main door and 5 meters away from the bathroom. He has difficulty negotiating the stairs in the house and in the school. The patient socializes with peers but his condition has hampered him from engaging in any sports activities.

On admission, the patient's vital signs were essentially normal. Pertinent physical examination findings were: height of 170 cm and weight of 50 kg with foreshortened left lower extremity, and true leg length measurement of 90 cm on the right and 52 cm on the left. The left hip was in a slightly flexed and externally rotated position. The left thigh was bulbous with a non-functional knee joint. The left foot was at a level just below the level of the right knee with the patient in standing position. There was no spinal deformity. There was atrophy of the muscles of the left posterior pelvis with no active range of motion (ROM) at the left hip and knee. Hip muscles were graded 0/5. There was full and pain-free passive ROM at the left hip and no appreciated motion at the knee joint. Normal ROM in all planes was noted at the left ankle with motor grade of 4/5. The patient presented with a stooped posture. The patient was independent in activities of daily living (ADL) but with difficulty in transfer activities, and ambulated using bilateral axillary crutches or by hopping on the right leg. The patient's baseline Functional Independence Measure (FIM) score was 121/126 with difficulty in transfers; a FIM score of 126 means complete independence. Radiographic study of the left hip revealed absence of the proximal portion of the left femur with dysplastic acetabulum and decreased bone density, anterior bowing of the left tibia and fibula (Figure 2) and normal left ankle and bones (Figure 3).

Rehabilitation problems identified were: significant leg length discrepancy of 38 cm; postural deviation; malrotation and flail left hip and non-functional knee; difficulty in transfer activities and ambulation; self-image issues; and non-participation in avocational (recreational) activities. Rehabilitation goals were set to correct the leg shortening non-surgically, stabilize the hip and compensate for the functional deficits, promote positive body image, and facilitate participation in sports. The pre-prosthetic rehabilitation program of the patient focused on strengthening of extremities and trunk, improving weight bearing on the shortened extremity, and improving transfer skills and ambulation using bilateral axillary crutches. The patient had difficulty in transfer activities and in ambulation requiring bilateral axillary crutches. The patient was observed to be timid during interviews and during the course of stay at the hospital. Psychological evaluation

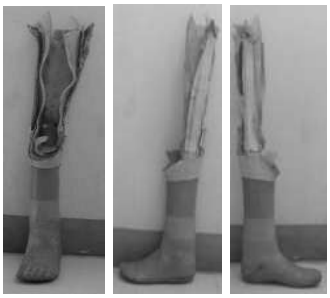


Figure 1. Extension prosthesis used by the patient from 4 to 8 years old.

Figure 2. Radiographic study of the left thigh and leg.

Figure 3. Radiographic studies of the left ankle and foot.

revealed no clinical signs of depression. The patient was discharged on the fourth hospital day with functional improvement. The patient was discharged trained on modified transfer technique and ambulatory using bilateral axillary crutches with correction of posture and gait deviation. He was provided a home program on continuing pre-prosthetic rehabilitation.

Five months later (October 2008), the patient was re-admitted and fitted with a "prosthosis". The patient underwent prosthetic rehabilitation to strengthen the trunk extensors, flexors and oblique muscles, right lower extremity, left ankle and left foot; to further improve transfer skills and ambulation using the prosthesis; and to address body image issues. The patient was prescribed with functional exercises in transfer activities and gait training with the "prosthosis" (level ground, ramp, uneven terrain, stairs). The patient was referred for psychological counseling to explore the patient's body image perception while on "prosthosis." Follow-up evaluation revealed acceptance and satisfaction with the prosthesis. He was discharged on the 10th hospital day, knowledgeable on prosthesis care, independent in prosthesis donning and doffing, ambulatory with the prosthesis with minimal trunk listing during stance phase on the prosthesis side without assistive device. His FIM score improved from 121/126 to 124/126. He was provided with a prosthetic rehabilitation home program and was advised to have regular out-patient visits for monitoring. On follow-up in March 2009, the patient reported continued use of the prosthesis and participation in basketball games with peers which he had not been able to do since he was eight years old. He also showed more confidence during conversation. The patient's most recent follow-up was in March 2010 for length adjustment of the prosthesis. He was in college at the time of follow-up.

Discussion

In patients with PFFD, the proximal femur is partially absent and the entire limb is shortened. Biomechanical abnormalities include limb length discrepancy, malrotation, proximal joint instability, and inadequacy of the proximal musculature. There is no identifiable etiology in this patient.

The most common and widely used classification to describe congenital femoral anomalies is the Aitken classification which divides PFFD into four categories based on the radiographic appearance.[5] This patient is classified as having class C with radiographic findings of absence of the head, neck, trochanter and middle portion of the left femur, and dysplastic acetabulum.[5]

Current treatment options are: use of prosthesis without surgical correction which includes Moseley Device (Figure 4) and hybrid prosthesis (Figure 5); use of prosthesis with surgical correction (arthrodesis of the knee without ablation of the foot) and prosthesis (Figure 6); arthrodesis of the knee with ablation of the foot and prosthesis (Figure 7); Van Ness rotationplasty and prosthesis (Figure 8); and limb lengthening (Figure 9).

In the case study by Moseley of a patient with PFFD, he illustrated the following advantages of his device and unrotated ankle (Figure 4): 1) no disruption of the stability of the knee; 2) adequate body awareness because of intact proprioception; 3) spontaneity of use of unrotated ankle acting as the knee joint function, simulating the normal gait; and 4) maintenance of intact body image.[6]

Hybrid prosthesis combines orthotics and prosthetics technology; as such, these devices are called "prosthoses" (Figure 5). "By using a thermoplastic solid-ankle-foot orthosis system that is attached distally to a prosthetic-type pylon and foot plate, the patient can easily control the ground-reaction forces which are more optimally transferred to the extremity. This enhances not only stability, but also gait speed and agility.[7]" Adjustments can be made to lengthen the device and accommodate the growth to maximize its use. A patient's enhanced proprioception and muscle use facilitate the development of a more normal gait.[7]

On the other hand, surgery with the use of prosthesis includes arthrodesis of the knee without foot ablation (Figure 6) and with foot ablation (Figure 7). In both procedures, the advantages are similar with Moseley Device. In addition, flexion of the anatomical knee within the socket of the prosthesis is now eliminated. In arthrodesis of the knee with foot ablation, an end-bearing stump is created and the patient is fitted with a prosthesis as a transfemoral (above-knee) amputee (Figure 7). This is currently the most commonly used treatment.[8] The advantages of this procedure are the creation of a good end-bearing stump with self-suspending potential and a much easier prosthetic fit. The disadvantage is a decreased ability to ambulate without a prosthetic device.[6]

In Van Ness rotationplasty, the foot is rotated to 180 degrees so that the ankle joint functions as a knee joint (Figure 8). The rotated foot becomes the transtibial (below-knee) stump for the below-knee prosthesis. The main prerequisites for the Van Ness procedure include unilateral involvement, a stable hip, presence of the fibula, a normal foot, and active ankle ROM of at least 45 degrees of plantarflexion.[9] The primary drawback of this operation is cosmesis as the foot is pointing backward, hence poor psychological acceptance by patients especially when the patient is female.[10] There is also a tendency for the ankle to derotate, which may require revision during the child's growth period.[11]

Limb lengthening was not an option for this patient because of the significant leg length difference of 32 cm (Figure 9).[12] Limb lengthening is a multi-stage process that is usually started before the age of three, and optimally completed by high school age.[13] Indications for lengthening include a limb with a predicted discrepancy at maturity not exceeding 20 cm, a hip that is or can be made stable, and a

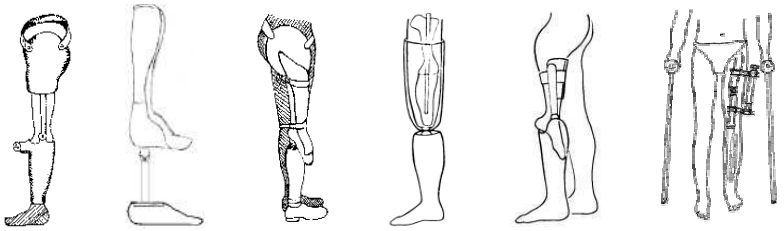


Figure 4 Figure 5 Figure 6 Figure 7 Figure 8 Figure 9

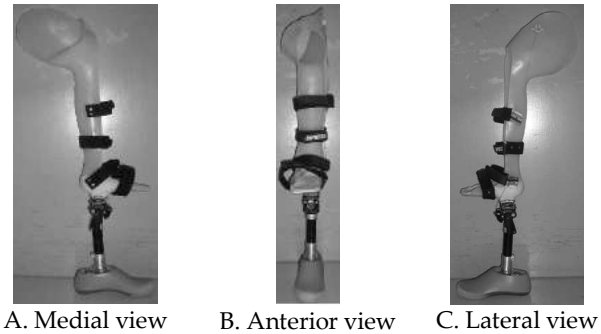
Figure 4. Moseley Device[6]
Figure 5. Hybrid prosthesis[7]
Figure 6. Use of prosthesis with surgical correction (arthrodesis of the knee without ablation of the foot) and prosthesis[6]
Figure 7. Arthrodesis of the knee with ablation of the foot and prosthesis[6]
Figure 8. Van Ness rotationplasty and prosthesis[6]
Figure 9. Limb lengthening[12]

relatively good knee, ankle, and foot. Maximum lengthening should not exceed 8 to 12 cm in older children and 4 to 6 cm in younger children. Before lengthening can begin, it is important to first stabilize the hip joint through arthrodesis. Without this, there is an increased risk for dislocation of the hip joint during lengthening. The patient will require one or more femoral lengthening and/or contralateral epiphysiodesis.[14] Since the patient has not reached his full skeletal maturity, a surgical procedure was not favored; thus, a prosthetic device was especially designed to address the significant leg length discrepancy and underdeveloped musculature (Figure 10).

The design used for our patient is similar to Figure 5 with modifications. The thermoplastic solid hip-ankle-foot orthosis was used to accommodate and provide stability to the shortened left leg. The orthosis also served as the proximal attachment for the standard transtibial (below-knee) endoskeletal prosthesis designed with a mechanical hinge joint and a manual locking mechanism using a cable attached to the lateral proximal part of the thigh shell.

The mechanical hinge acts as the "knee" that allows the device to flex when the patient sits (Figures 11 and 12). However, cosmesis remains a problem. There is protrusion of the patient's anatomical foot during sitting (Figure 12). The mechanical hinge joint is locked in extension during ambulation; gait deviation including absence of mechanical hinge "knee" joint flexion during the swing phase of the prosthetic leg and listing of the trunk toward the prosthetic side during the stance phase was unavoidable (Figure 13). Issues regarding body image and gait deviation were addressed through comprehensive, holistic rehabilitation. The Prosthesis Evaluation Questionnaire (PEQ) is a validated tool to assess various aspects of prosthesis use; this was administered to the patient. The PEQ uses a visual analog scale with 0 mm as "extremely unhappy or terrible" at one end and 100 mm as "extremely happy or satisfied" at the other end. Our patient rated "extremely satisfied" with his ambulation, appearance and utility of the prosthesis, perceived response of strangers, and functional outcome changes in his quality of life.

Although the prescribed "prosthosis" did not correct the structural deformity, the device enabled the patient to compensate for his functional deficits in transfers and ambulation, and reduced his instability during standing and independent ambulation. Consequently, prosthesis use improved the patient's self-confidence and allowed him to engage in basketball as an avocational pursuit. The patient continued to use the prosthesis with regular length adjustment of the endoskeletal shank to accommodate increases in height during the succeeding two years.



A. Medial view B. Anterior view C. Lateral view

Figure 10. The prescribed "prosthosis"

Figure 11. Patient performing sitting and standing activities with "prosthosis"

Figure 12. Position of the anatomical foot when patient is seated.

Figure 13. Independent ambulation with gait deviation.

Individuals with PFFD have normal life expectancy. It only took one "prosthosis" to transform our patient form being a shy teenager with some disabilities and handicaps into a functionally independent young man despite being differently abled.

Conclusion and Recommendation

The management of patients with PFFD requires a multidisciplinary team approach involving the rehabilitation medicine specialist (physiatrist), pediatrician, orthopedic surgeon, prosthetist, orthotist, physical therapist, psychologist and other specialists depending on the changing needs of the patient. The primary goal of treatment is to optimize function especially ambulation. The prescribed device is a cost-effective, non-surgical alternative for patients with PFFD. The major benefits of using this prosthesis are: safe mobility, independent ambulation, and participation in recreational activities. Prosthesis use also promotes positive self-image. Regular follow-up on long-term basis is recommended to monitor mechanical breakdown of the prosthesis, to provide adjustments to accommodate continuing growth, and to assess compliance to prosthesis use and effectiveness of the prosthesis. Monitoring should also include anticipatory management of potential musculoskeletal complications such as scoliosis, pain and skin breakdown, and emerging psychosocial concerns of the patient.

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