

# Late Referred Proximal Focal Deficiency: A Comprehensive Rehabilitation Management - A Case Report

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**Abstract:** A child with Proximal Focal Femoral Deficiency (PFFD) would suffer a gait problem due to leg deformities. PFFD is a rare congenital anomaly ranging from one case per 50,000 population to one case per 200,000 population. This is a case report of a 5 years old girl with isolated left PFFD Aitken D. She met the orthopedic at the age of 6 months and asked to come back at 5 years. She has left knee contracture and limited involved hip range of motion also hypermobility joint in the uninvolved leg. The patient walking by kneeling to compensate for the leg length discrepancy (LLD) and has an inadequate function of the uninvolved leg. The rehabilitation program consists of a home-based strengthening and stretching exercise program, gait training, balance exercise, elastic tape application, and an adaptive prosthesis. At the end of 2 months of comprehensive rehabilitation management, the balance has improved and the patient was able to stand at the same level as her friends and walking with an adaptive prosthesis with a better gait pattern.

## 1 INTRODUCTION

Proximal focal femoral deficiency (PFFD) is a congenital anomaly that involves the pelvis and proximal femur with widely variable clinical manifestations, from mild femoral shortening and hypoplasia to the absence of any functional femur and acetabulum aplasia (Dillon et al., 2005). The more severe presentations would have progressive subluxation to dislocation of the femur (Alexander et al, 2015). Aitken in 1969 made a simple classification for PFFD which is widely used worldwide. This divides PFFD into four classes based on the radiographic appearance and the anatomic relationship between the acetabulum and the proximal end of the femur (Lasanianos and Kanakaris, 2015). The child may need a socket at a relatively young age to capture the limb and provide maximum stability. Surgery is not the first option for PFFD treatment, it's indicated to made more acceptable stump, provide hip stability and improving gait pattern when wearing a prosthesis (Alexander et al, 2015). The fusion of the femoral

remnant to the tibial remnant would effectively create a residual limb that functions and has prosthetic needs of a knee disarticulation patient. A Van Ness rotationplasty is another surgical procedure frequently used in this case (Ackman et al., 2013).

## 2 CASE REPORT

A 5 years old girl came to our hospital in the last week of July 2019 with PFFD Aitken D. She lived in a small village in Pasuruan district, about 90 km from Surabaya. Her father was a carpenter, working on a local businessman, while her mother was an honorary clerk at the restaurant near the house. She started kindergarten this semester. She is the only child, spontaneous delivery at term by a midwife, with 3700 grams birth weight and spontaneously crying. No clear cause at prenatal history. At 6 months old she has met the orthopedic, got X-Rays and come back at the age of 5 or 6 years old. Her development was similar to another child, she eats

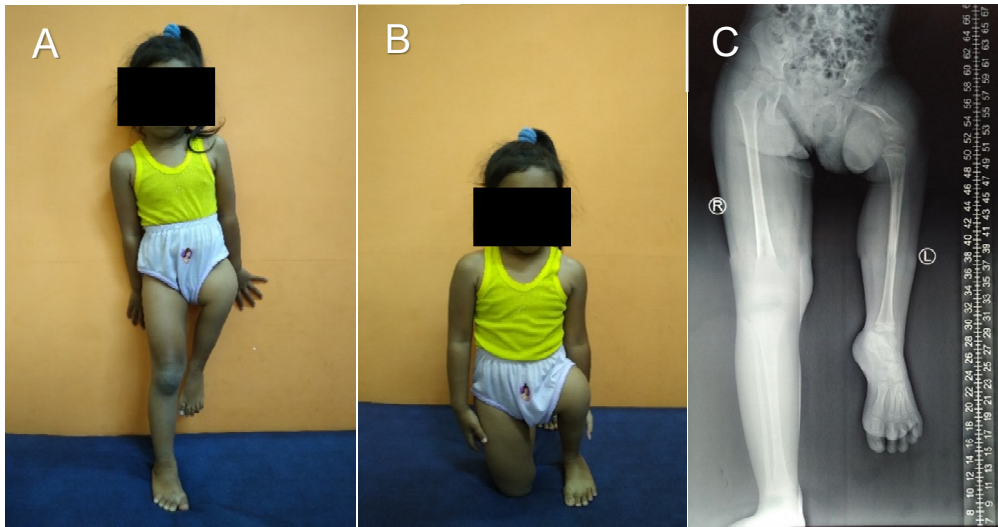


Figure 1: Clinical appearance patient. (A) The front view, the left ankle at the right knee level. The girl can not stand without holding to something. (B) Patient position when walking indoor, the right knee touching the floor with her lower leg bending backward and the left ankle in tip toeing. (C) Radiograph image. The left acetabulum not formed and absence of femoral head. The size of the left femoral shaft is less than 50% of the right femur. Note the linear blurry image, which caused by patient's movement due to poor balance.

the same food as the family, communicate verbally, but shy with strangers. She could write a letter. No complaint on pee nor defecation. Based on her mother, the patient could walk as far as about 300 m to play to the neighbors. The girl wants to stand upright and joint the carnival to celebrating the independent day.

On examination, we found the left thigh was much shorter than the right one, whereas the size of the tibia and foot at both sides were the same (Fig 1). The body weight 16 kg with body length 102 cm, and head circumference 102 cm. She had limitations range of movement (ROM) on her left hip, the left knee ROM was 60 – 135° with hard end feel and. The manual muscle test (MMT) of left hip flexor and extensor were 2. She can't do one leg stands (OLS) with open eyes on her good leg. The girl walked by kneeling her right knee to compensate for the difference in leg length. When in the house, she walks with kneeling the right knee, while on the ground, she walks using both of her foot so the right knee at her chest level. She used pelvic rotation to move forward to compensate for the weakness of the hip flexors. There were no anomalies were found on the skeletal, cardiac, gastrointestinal, genitourinary, or nervous systems were observed. No trauma she got, no pain, no swollen or redness on her left leg. There were no family members has the same deformity as hers. For the time being, the parent not considered surgery as

the acceptable treatment. The radiograph in standing position, show there were no femoral head nor acetabulum on the left side. The femoral remnant hanging in abduction and flexion position. (Figure 1C).

In the first week of rehabilitation program, our main goal was to increase her balance. Due to the socioeconomic condition, she was given a home-based exercise program. The program would be evaluated every 2 weeks. The progress sent weekly by the mother via Whatsapp®. We taught her mother to stretch the hip abductor, flexor and internal rotator, bridging exercise, sit to stand and how to walking with the walker, OLS practice and strengthening lower extremity. Continuous counseling also was given both to the patient and the parent to help them to accept the girl's condition and boost the girl's confidence. The adaptive prosthesis was made with specification: ischial containment adaptive socket with an open front and footrest, knee joint, shank, each foot (Figure 2A).

Two weeks later, the girl already could do OLS less than a second and could walk with a walker. We found the Lachman test, valgus and varus stress test on the right knee were positive. Anterior drawer test in both ankles was also positive. The hypermobility joints were not helped her balance. We gave her elastic tape on her right knee and ankle to provide more stability. Soon after elastic tape application, the girl could do OLS for four seconds. In the fourth

week, the girl could do OLS for 2 seconds without elastic tape, and six seconds with elastic tape. We give elastic tape once every 2 weeks which lasts for 5 days on the girl.

### 3 DISCUSSION

PFFD is an anomaly that results from the



Figure 2. (A) The adaptive prosthesis with adaptive front open ischial containment socket and foot rest. (B) The girl stand in her good leg. (C) The girl and her adaptive prosthesis anterior view. (D) Lateral view of the girl

The girl got adapted prosthesis at the sixth week of our rehabilitation program, but she rejected her prosthesis. She cried and would not look into it. We explained to the parents and showed the girl some videos with a girl wearing lower limb prostheses. We monitored as the girl slowly accept her prosthesis. She had difficulty to control the knee unit, so we suggest the parent to lock the knee momentarily. The girl's response was good. She felt more stable with the knee locked in the extended position. The next week, she could walk with the prostheses by handheld to the parents. At the eighth week, she already could walk some steps with her prosthesis independently, with fast steps and limping gait. There were no pain nor discomfort during the use of prosthesis and no redness nor blisters on the skin that contact with the socket. After given some correction on her gait and motor education, she could walk further (about 5m) independently (Fig 2 B,C). The pelvic rotation is reduced. The balance was significantly improved, she could do OLS for 7 seconds without elastic tape. However, the girl's still couldn't do hopping and sit to stand without hold. She still has to hold on when walking long distance. The strengthening exercise continued at home and the gait exercise would be done every 2 weeks.

development failure of the subtrochanteric portion of the femoral shaft. It's a rare skeletal disorder ranging from one case per 50,000 population to one case per 200,000 population (Lasanianos and Kanakaris, 2015). It could be diagnosed early at 20 weeks gestational age using ultrasound with characteristic the absent of femoral head and shortening femoral shaft (D'Ambrosio *et al.*, 2016). Early diagnosed PFFD would help the parent to accept their child's condition. Although the etiology of PFFD is uncertain, the common etiologies of the short femur should be excluded, such as aneuploidy (especially, trisomy 21), poor diabetic control, exposure to drugs (thalidomide), viral infections, radiation, focal ischemia between the 4<sup>th</sup> and 8<sup>th</sup> week of gestation (Lin *et al.*, 2013).

Aitken in 1969 has described a classification that has been widely used. The radiographic findings are ranked from a benign form (A) to a severe form (D) according to the extent of the femoral deficiency. Type A has a small area of deficient femur below the femoral head. In type B this same segment is absent. In type C the femoral head is absent and the acetabulum deformed. Finally in type D both the acetabulum and femoral head are absent (Lasanianos and Kanakaris, 2015).

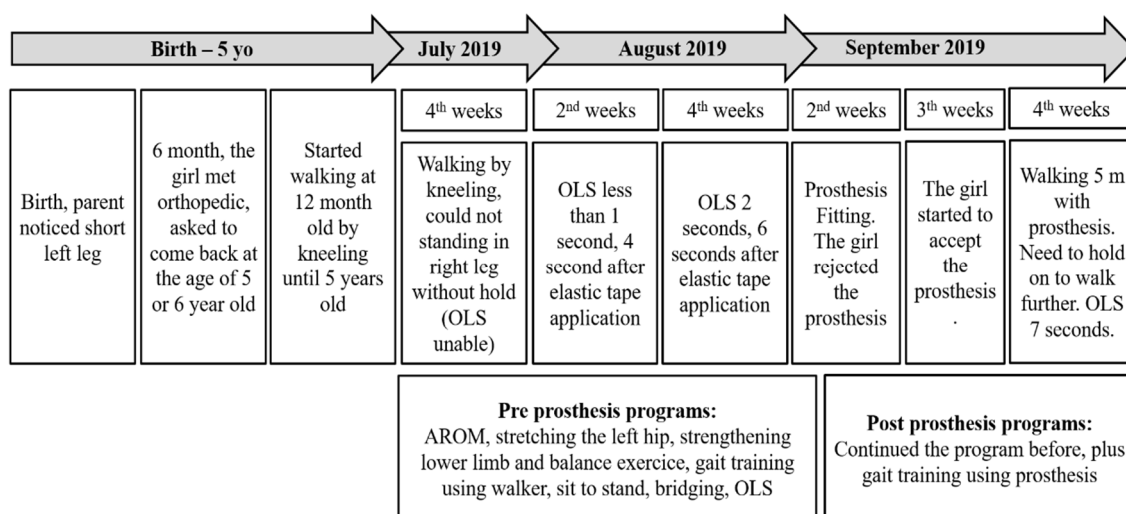


Figure 3: The timeline of the patient's history. OLS: one leg stand. AROM: active range of motion

Each child with PFFD must be assessed individually since no single treatment approach applies to all cases. Limb lengthening is indicated in cases which predicted discrepancies at maturity not exceeding 20 cm, in hips that are or can become stable and when a relative good knee, ankle, and foot is present. In the case of predicted discrepancies greater than 20 cm or if other reasons do not render a child suitable for limb lengthening, prostheses should be considered. Treatment options could be generalized based on Aitken classification which vary from shoe lift, prosthesis, and both surgery and prosthesis. In classes C and D a stable hip joint cannot be obtained thus a fusion of the hip and, possibly, the knee joint is necessary (Lasanianos, Kanakaris and Giannoudis, 2015). The fusion of the femoral remnant to the tibial remnant, would effectively create a residual limb that functions and has prosthetic needs of a knee disarticulation patient. A Van Ness rotationplasty is another surgical procedure frequently used in this case (Ackman *et al.*, 2013). A Van Nesh rotationplasty indicated when the ankle level of the affected side is at the same with the knee level of the unaffected side. When the femoral remnant is less than 50%, it would made the ankle fell above the knee, Syme amputation is the recommended procedure to provide a good stump. However, surgery not the first option in this case. Since the girl's parent objection for the surgery and the girl already could walk by herself, we decide prosthesis would be the best treatment choice for the time being.

Weakness and contracture of the left limb of the girl due to lack of use. She walks in an abnormal position for her first 4 years. As the prosthesis use

needs several qualifications, exercise is also the main treatment in this case. Side lying in the left side would stretch the hip abductor, bridging would target hip extensor, and sit to stand exercise would activate the quadriceps muscle and increased knee stability (Da Costa, Savelsbergh and Rocha, 2010). The child with lower limb deficiency should be fitted with a prosthesis when he or she is ready to pull up to a standing position (Abudu *et al.*, 2006). This usually occurs between 9 and 10 months of age. The goals in fitting a prosthesis at this early age are to allow for normal two-legged standing, proprioceptive input, provide a better means for reciprocating gait development, and provide a normal appearance (Alexander et al, 2015). The prosthesis should be simple in design, allow growth adjustment, suspend securely, and preferably lightweight. It is very important to establish a standing position for the girl as soon as possible. It would provide better proprioceptive which is important for walking endgram (Barra and Pérennou, 2013). The gait pattern maturity around the age of 7-8 years old. As the girl is now 5 years old, it is important to improve the child's posture and walking pattern at this time.

The girl has hypermobility syndrome, which worsening her balance. We put elastic tape on her right knee and ankle so it would help to stabilize the leg. Elastic taping could give the proprioceptive stimuli to establish the joints (Chinn *et al.*, 2014). On the second visit, elastic tape application was significantly improved OLS time, from 1 second to 4 seconds after elastic tape application.

The home-based exercise is more often used in elderly patients with frailty (Clegg *et al.*, 2014). In

this case, due to the socioeconomic conditions of the patient and the location of the house which is quite far from our hospital, we thought home-based exercise program is the best option for the patient. In this case, the result of home-based exercise mostly depends on parent's contribution. Adequate education must be given. Prosthesis rejection also became some issue in our rehabilitation program. It took a week for the girl to accept her prosthesis and started the gait exercise using a prosthesis. Postema et al recommended the age of the first fitting for upper-limb prosthesis, not more than 25 months. Fitting after this age seems to be related to higher rejection rates. The other systematic review by Meurs et al said that fitting age of prosthesis depends on clinical experience rather than evidence-based (Meurs *et al.*, 2006). The girl had difficulty to control the knee unit given in the adaptive prosthesis. The locked knee in full extension helped the girl to walk better. The knee unit in children's first prosthesis rather difficult to control (Cummings, 2000). As the girl's control of her prosthesis getting better, the knee unit could be used. The myoelectric prosthesis would be the best choice for this condition (Tsukishiro, 2003). However, it would be cost too much for the family.

For the future, the girl still needs to be routinely evaluated. We continued to monitor for any redness or blisters due to the use of prostheses. As the girl continues to grow, the prosthesis would need several times to adjust. The exercise should be done regularly. It is important to improve ROM and strength of both of the legs. The continuous counseling for both the child and the parents would be need.

## 4 CONCLUSIONS

It is important to know the characteristic of PFFD and the treatment option we could offer to the patient. Treatment of PFFD not as simple as to choose between either prosthesis or surgery. We need a comprehensive rehabilitation program that is tailored as the patients need. We suggest the patients with PFFD came to a physiatrist soon after birth.

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