

# Prosthetic Restoration for Bilateral Congenital Lower Limb Transverse Deficiency

## Bilateral Konjenital Alt Ekstremitte Transvers Gelişim Anomalisinde Protez Uygulaması

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### ABSTRACT

Congenital transverse deficiency of lower limb is a rare defect, which may cause difficulties in ambulation, daily living activities and self care activities. Suitable prosthetic restoration is an important step in rehabilitation of such patients. In this case we present our approach of lower limb prosthetic restoration to a patient with deficiencies at different levels in both lower extremities.

**Keywords:** Congenital, limb, deficiency, prosthetics, rehabilitation

### ÖZET

Alt ekstremitenin konjenital transvers anomalisi nadir görülen ve hastanın ambulasyonu, günlük yaşam ve kendine bakım aktivitelerini olumsuz yönde etkileyen bir durumdur. Uygun olan en erken zaman diliminde protez uygulanması bu hastaların rehabilitasyonunda önemli bir basamaktır. Bu olguda her iki alt ekstremitede, farklı seviyelerde transvers anomalisi olan bir hastaya yönelik protez uygulaması sunulmaktadır.

**Anahtar sözcükler:** Konjenital, ekstremitte, anomali, protez, rehabilitasyon

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## Introduction

Congenital transverse deficiency is a development failure with complete absence in a limb distal to some point, ending with a distal bud (1,2). It is also known as congenital amputation, failure of development, transverse melia, transverse arrest or terminal transverse absence.

Most congenital amputations have no particular established cause however amniotic band constriction, familial assendence and injury and development failures in first 6 weeks of pregnancy are some factors thought to take part in etiology. (3).

Prevalance of congenital transverse deficiency is found to be 19/100000(4). In most cases defect is unilateral. 40 percent of the children with congenital limb deficiencies will have multiple limb involvement, such as combined upper and lower extremity deficits. (3) Patients with such defects may suffer from many difficulties primarily in ambulation, daily living activities and self care activities, therefore prosthetic restoration is an important first step in the rehabilitation procedure.

In this report, a case with transverse arrest in left lower extremity and right foot and in both hands and our prosthetic restoration for lower limbs is presented with the approval of her parents.

## Case presentation

A two-year-old Caucasian girl presented to our clinic by her parents, for determination of suitable prosthetics as she suffered from lack of left lower extremity after knee joint, right foot after heel, and fingers in both hands congenitally.

In physical examination patient was oriented, cooperated, and there was no sign of mental retardation. Neurological examination was normal, patient was able to sit, crawl but unable to walk due to her condition.

Clinic and radiologic examination revealed transverse deficiencies below knee in left leg and distally to talus and calcaneus in right foot (Figure 1a,b,c). In upper limbs development was normal till the end of radius and ulna, there were radial epiphyses in both hands but no ulnar epiphysis and carpal, metacarpal bones and phalanges were absent, yet there were few rudimentary bone formations in both hands. Patient also suffered from an oropharyngeal malformation without any significant effect on speaking or swallowing. Patient was followed by plastic and reconstructive surgery for hand and oropharyngeal malformations.

Questioning of parents for patient history revealed that patient is the fourth living sibling – neither of them has similar condition – of mother's five gestations. Pregnancy was followed up for five months during which no abnormality detected with ultrasonography. Mother stopped attending to follow-ups after fifth month. Patient was given birth in ninth month of gestation by spontaneous vaginal delivery. Birth weight was 2750 gr. Patient history also revealed that there was second degree relation (cousins) between parents. There was no history of using any kind of prosthetics, orthotics, helping devices or attending rehabilitation before.

In order to provide ambulation, below knee socket prosthesis, with Y-band for left limb and promalleolar, partial foot socket prosthesis for the right limb, made from thermoplastic material were provided (Figure 2). This restoration with basic foot and ankle components was planned to be temporary as the patient had never walked before and the leg length and stump size may increase by age. Patient was successful in erect posture with the support of one parent. As the patient lived in a different city, home-based exercise programme was taught to parents and prosthetics education under supervision was also recommended in addition to regular follow-ups.

In first follow-up after 2 months the patient was able to walk with support of one parent. As the patient had



**Figure 1:** a: View of lower extremities of the patient.  
b: X-ray of right foot.  
c: X-ray of both knees.

overgrown her prosthetics, new prosthetics with same features were reconstructed. In follow-up after 6 months it was observed that patient was able to walk without any support.



Figure 2. View of the patient after prosthetic restoration.

## Discussion

Congenital transverse deficiency of lower limb is a rare defect, and rehabilitation and prosthetic restoration should be patient specific as deficiencies may vary. Most of the patients with congenital limb deficiencies are treated under age 15, 10% of which have congenital anomalies that are treated as, or require, amputation. However most transverse deficiencies are homologues of acquired amputations and can receive prosthetic evaluation and fitting as such (3).

Age, staging and growth should be taken into consideration while prosthetic treatment is being planned. Pulling up to stand generally occurs 9-12 months of age which should be the time of fitting prosthetics. Independent ambulation expected to begin between 15 – 22 months. (4) Therefore providing prosthetic restoration at early ages may be beneficial for patients with congenital deformities to avoid delay in functional development. Although the usage of prosthetics before crawling is disputed, after crawling is achieved by the child, prosthetic restoration is definitely needed in order to advance from crawling to standing as it is in our case (5). Children born with congenitally limb deformities also adapt well to the loss of function and manipulate prostheses as they tend to develop compensatory mechanisms easily (6). Prostheses need to be replaced every 15–18 months on the growing child. Some children may require a new prosthesis every year until age 5, then every 2 years between 5–12 years, and every 3–4 years until adulthood. (7)

We present this case in order to provide an example for management of similar cases as there is small amount of reports in the literature about this subject to our best knowledge, and to emphasize the importance of early prosthetic restoration.

The objective of providing partial foot prosthesis and below the knee socket prosthesis in this case is not only to prepare the patient for adaptation to using higher functioning prosthetics in the future, but also to provide ambulation and make the patient less dependent in daily activities and in mobilization as well. With proper rehabilitation, prosthetics education and patient participation, which assumed to increase by age, ambulation level is expected to increase in the future. With the increase in activity and ambulation level, using higher functioning prosthetic materials may be taken into consideration in order to optimize walking.(4)

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