

# Clinical Quiz

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## A child with congenital amputation of the foot

### Clinical Presentation

Congenital amputation of a limb is a rare birth defect characterized by failure of formation of the terminal part of an extremity. Also known as congenital transverse deficiency, this condition is thought to be due to mechanical disruption of a developed limb rather than a congenital malformation.<sup>1</sup> It can affect one or more limbs<sup>2</sup> and an upper limb deformity is more commonly seen.<sup>3</sup> We present here a case of congenital amputation of foot as clinical quiz to briefly highlight the clinical features of this rare birth defect and its etiology and management.

A 2-month old male child was brought to our outpatient department with the complaints of absent left foot and toes since birth. He was the first child of non-consanguineous parents born at term by an uneventful vaginal delivery. Although the mother did not attend regular antenatal checkups, there was no history of fever with rashes, drug intake or exposure to radiation during pregnancy. Clinical examination revealed absent forefoot and midfoot with rudimentary toe buds. It looked like a surgical partial foot amputation (**Figure 1**). There was no evidence of congenital constriction bands or limb length discrepancy, and rest of the skeletal survey was normal. Examination of the chest, abdomen and cardiovascular system were normal. Radiographic examination showed normal tibia and fibula with absence of all the bones of left foot except talus and few rudimentary bones (**Figure 2**).



**Figure 1** - Clinical photograph of the child showing partial absence of left foot with intact plantar fat pad (arrow head) and rudimentary toe buds (thin arrow). Right foot is normally formed.



**Figure 2** - Antero-posterior radiograph of both feet showing absence of all the bones of left foot except talus (large arrow). Few rudimentary bones are also visible (small arrow). Right side is absolutely normal.

### Questions

1. What is the diagnosis?
2. What is the etiology of this condition?
3. What is the management?

# Clinical Quiz

## Answers

1. The child has congenital amputation of left foot (congenital Chopart's amputation) classified as congenital transverse, tarsal, partial limb deficiency as per International Society for Orthotics (ISO) and International Society for Prosthetics and Orthotics (ISPO) classification.
2. The exact etiology of congenital limb deficiency in most cases is not known. Many teratogenic drugs like thalidomide have also been implicated in the etiology of congenital limb deficiencies.<sup>1</sup> However, majority of limb anomalies occur sporadically without any obvious traumatic, genetic or environmental factors.<sup>4</sup> Congenital amputation is thought to be due to mechanical disruption of a developed limb rather than a congenital malformation. The suggested mechanical factors are amniotic bands (Streeter band syndrome or congenital constriction band syndrome). At present, the most common cause of congenital foot deficiencies is believed to be the constriction band formation.<sup>1</sup>
3. Children with congenital partial foot deficiency usually do not require surgical treatment and have minimal, if any, functional limitations. They can walk and can even climb stairs like normal children.<sup>5</sup> A child with Chopart foot can walk bearing full weight on the affected limb. This is made possible because of maintenance of functional limb length and stump having broad weight bearing surface with preserved heel pad.<sup>1</sup> A slipper-type prosthesis (shoe filler) can allow normal shoe wear and function.<sup>1</sup> When the foot is too short to suspend prosthesis, ankle foot orthosis with a foot plate and shoe filler may be used. Along with the prosthetic management, it is important to encourage normal activities and psychological build up for these children.<sup>4</sup>

## Discussion

Congenital limb deficiency is not a common problem. Upper limbs are involved more commonly than the lower limb.<sup>3</sup> These deficiencies are broadly classified by ISO and ISPO as either transverse or longitudinal. Missing bones are named and described as either complete or partial in their absence. In congenital transverse deficiency, formation of parts of a limb does not occur distal to certain level leading to total absence of terminal part. The resulting limb looks like an amputation stump. It has differently been described in literature by terms like congenital amputation, terminal absence, transverse melia, transverse arrest or terminal transverse absence.<sup>3</sup>

Congenital absence of foot is an uncommon anomaly and this has further been classified into congenital Chopart's (transverse, tarsal, partial), congenital absence of forefoot (transverse, metatarsal, complete) and congenital Syme's (transverse, tarsal, complete).<sup>4</sup> Our case was of hind foot amputation classified as congenital transverse, tarsal, partial deficiency as a well formed talus was present. Jain and Lakhtakia<sup>3</sup> found 4 cases of congenital amputation of foot in their series of 36 cases. They excluded amputation through digits and toes from the study. They observed that the transverse deficiency at the level of hind foot was less frequent than those at the forefoot. Unilateral as well as bilateral congenital amputations were more frequently seen in the upper extremity as compared to lower extremity. Although most common cause of congenital foot deficiency is said to be constriction band formation (streeter's bands),<sup>1</sup> no evidence of any constriction band was present in our patient. The goals of the management of these children with foot deficiencies are to maximize the child's functional independence and minimize the psychological impact of the condition. Early prosthetic fitting in a child with congenital amputation may be beneficial to prevent delay in functional development. This helps in easy adaptation to the deformity and development of good compensatory mechanisms. A growing patient requires frequent change of prosthesis usually every 15-18 months.<sup>2</sup> As discussed above, these children do very well functionally with prosthetic management in the form of a slipper-type prosthesis which allows normal shoe wear. But, the absence of foot may have a significant negative effect on the self esteem of the child. Children's understanding of their disability is incomplete at 6 years of age but at around the age of 8 or 9 years, they come to a much more complete understanding of their handicap.<sup>4</sup> So the older children and adolescents need encouragement for their psychological build up to cope with their disability.

In conclusion, we describe an uncommon and peculiar case of congenital amputation of foot at the level of hind foot. Application of slipper-type prosthesis along with psychological build up of the child results in good functional outcome. We feel that further studies by specialized paediatric orthopaedic units are required to know the actual incidence and etiology of this rare condition.

## References

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