# Lower limb deficiency syndromes

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## Abstract

Lower limb deficiency syndromes consist a spectrum of deformities which include undergrowth, overgrowth, failure of formation, failure of differentiation, duplication and constriction band syndromes. This article concentrates on the more common lower limb deficiency syndromes and their orthopaedic management.

**Keywords** congenital femoral deficiency; constriction band syndrome; fibular hemimelia; tibial hemimelia

## Introduction

Lower limb deficiency syndromes include a constellation of congenital abnormalities varying in severity from the complete absence of one or more limbs (with or without upper limb involvement) to hypoplasia of a limb segment or absence of a digit. The incidence of lower limb reduction defects is estimated to be 2 per 10,000 live births in the USA, although there is likely to be significant geographical variation and the true incidence in the developing world is not known.[1]

## Classification

Multiple classification systems exist to describe both upper and lower limb deformities. These can be descriptive, aetiological, anatomical or a combination. One of the most well-known classification systems for limb deficiencies is the Swanson classification, which can be applied to either the upper or lower limb. It is a descriptive classification based upon the anatomical location of the deficiency, the mode of embryological failure and whether there is total involvement (bone and soft tissue) or just dermomyofascial involvement (Table 1).[2]

The ISO/ISPO (International Organization for Standardization/International Society for Prosthetics and Orthotics) classification system is limited to deficiencies which are a failure of formation and describes them on an anatomical and radiological basis only. Deficiencies are described as transverse when the limb has developed normally up to a certain point, but beyond which there are no skeletal elements (although there may be vestigial digits). They are named by describing the limb segment where termination occurs and then the level within that segment, e.g. transverse leg, upper third. Longitudinal deficiencies describe reduction or absence along the long axis of the limb. The nomenclature for this is more complex. The affected bones are named and it is then stated if they are totally or partially absent. An estimate of the location and amount of bone absent is made, as well as the absence of any rays, e.g. fibula total, tarsus partial, ray 5 total.[3]

More recently Gold et al have described a classification system which encompasses both the anatomical abnormality and the aetiological pathogenesis of the limb deformity.[4] This system allows for a more accurate method of assessing overall prevalence rates as well as identifying phenotypes which are less well-known (see Table 2).

These classification systems are useful for categorizing patients on a population basis, although they have limited value to the practicing orthopaedic surgeon, as they do not guide treatment. Consequently, several condition-specific classification systems have been developed, which aid in decision making. These will be described in more detail later in this article.

## Embryology

The embryological development of the limbs occurs during weeks 4–8 following fertilization. It is during this window of time that the majority of congenital limb abnormalities initiate. The limb buds develop from mesoderm and ectoderm. The mesoderm forms the muscle of the limb and the ectoderm forms the bone and cartilage. The apical ectodermal ridge (AER) develops at the distal part of the limb bud as a thickening of the ectoderm and is involved in controlling the proximal to distal development of the mesoderm. In addition the AER is responsible for regulating apoptosis of the web spaces between the digits. If it does not function to do this, then webbing or syndactyly can occur. If the AER is absent then limb bud outgrowth is arrested and transverse arrest results. Additional AERs will lead to limb duplication.

The second stage of limb development involves extending the length of the limb via chondrocyte proliferation, followed by bone formation via the process of endochondral ossification. These processes are regulated by Hox genes. Defects in these genes lead to severe shortening and growth retardation.[5]

## Fibular hemimelia

Fibular hemimelia, although rare with an incidence of 10–20 cases per million live births, is the commonest deficiency of the lower limb (Figure 1).[6] It can be considered as a longitudinal,

### Swanson classification

| Type | Description | Example |
|---|---|---|
| 1 | Failure of formation | Transverse/longitudinal |
| 2 | Failure of differentiation | Soft tissue/skeletal/tumours |
| 3 | Duplication | Polydactyly |
| 4 | Overgrowth | Hemihypertrophy |
| 5 | Undergrowth | Brachysyndactyly |
| 6 | Constriction band syndromes | |
| 7 | Generalized abnormalities | |

**Table 1**

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**Gold et al. classification of congenital limb deficiencies**

| Aetiology or pathogenesis of limb deficiency | Example |
|---|---|
| Chromosomal abnormalities | Trisomy 18 |
| Known syndrome | VACTERL |
| Mendelian inheritance | Split hand/foot syndrome |
| Familial inheritance | |
| Presumed vascular disruption defects | Amniotic band syndrome |
| Teratogenic exposure | Misoprostol |
| Unknown cause | |

From Gold NB, Westgate M-N, Holmes LB. Anatomic and etiological classification of congenital limb deficiencies. Am J Med Genet 2011; 155 Part A: 1225–35.

**Table 2**

post-axial deficiency because it affects the long axis of the leg on the side opposite the great toe.[7] Although classically described as a partial or complete absence of the fibula, the deformity is more widespread throughout the limb, with varying severity of associated deficiencies, including: hypoplastic acetabulum, congenital femoral deficiency, hypoplastic lateral femoral condyle, genu valgum, anterior and often posterior cruciate deficient knee, shortened tibia with, classically, an anteromedial bow, valgus distal tibia, ball and socket ankle joint, tarsal coalitions (often of the hindfoot), equinovalgus (occasionally equinovarus) foot and absent lateral rays. The condition may be associated with upper limb abnormalities such as syndactyly, as well as the post-axial equivalent in the upper limb; ulnar hemimelia.

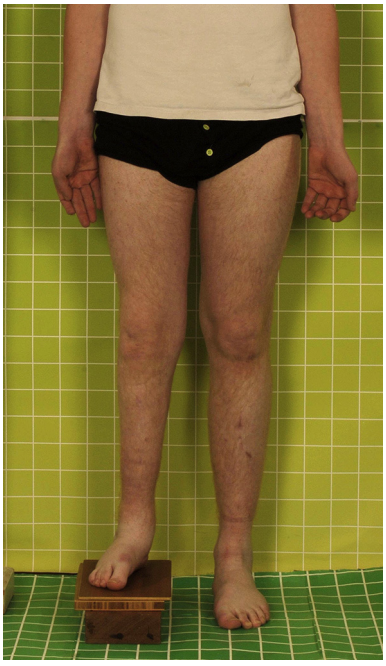

**Figure 1** Clinical photograph of an adolescent with fibular hemimelia, a four ray foot and residual shortening after previous lengthening.

There is significant variation in the severity of fibula hemimelia and, as such, there is significant potential for variation in treatment options. For the mildest cases, guided growth in the form of temporary epiphysiodesis can be used to correct genu valgum, whilst epiphysiodesis can be employed to correct mild limb length discrepancies. In severe cases, the limb may not be reconstructible, in which case amputation may be offered. Indications for amputation are individual and need to take into account the personal and cultural beliefs of the family. However, if a plantigrade, stable, three-rayed foot cannot be achieved then amputation should be discussed with the family.

## Classification

Multiple classification systems have been developed to describe fibular hemimelia. One of the most popular classification systems is that of Achterman and Kalamchi. It is a descriptive classification based on the degree of involvement of the fibula. Type 1 encompasses incomplete fibular deficiency. In Type 1A the proximal fibular epiphysis is distal to the tibial epiphysis, while the distal fibular growth plate is proximal to the talar dome. In Type 1B 30%–50% of the proximal fibula is absent, while distally it is present but does not support the ankle. Type 2 encompasses complete fibular deficiency, or where only a distal vestigial fragment is present.[8] This classification system was published in 1979. At that time the authors recommended leg length equalization for Type 1 and amputation for Type 2. As understanding of this condition has evolved and technological and surgical techniques have improved, there is now less reliance on the presence or absence of the fibula in the decision making process for either amputation or reconstruction and more emphasis on the presence or absence of a functional foot. This is reflected in the classification system of Birch, who divided fibular hemimelia into two groups; Type 1 having a functional foot and Type 2 having a non-functional foot. Type 1 is subdivided into four sub-types according to the limb length difference with the unaffected side: 1A <6% difference, 1B 6–10% difference, 1C 11–30% difference and 1D >30% difference. Type 2 is subdivided into two groups: 2A functional upper extremity, 2B non-functional upper extremity.[9] This classification system aims to be both descriptive as well as potentially guiding the surgeon's treatment strategy.

Paley has also described a classification system, which aims to guide the surgeon on various reconstructive options depending upon the deformity.[10] It does not take into account the presence or absence of the fibula, nor does it comment on overall limb length discrepancy (Table 3).

## Treatment

In Paley Type 1 fibular hemimelia the principles are to address any Achilles tendon contracture with a soft tissue release and simultaneously lengthen the tibia whilst correcting any bowing deformity present. In Type 2 fibular hemimelia, where the ankle valgus is dynamic, the peroneal tendons can also be lengthened in addition to realigning the ankle joint with a supramalleolar osteotomy, either prior to, or simultaneously with the lengthening procedure. The equinovalgus deformity in Type 3 fibula hemimelia can be addressed either by a supramalleolar osteotomy if it originates from the ankle, an osteotomy at the site of the subtalar coalition if it arises below the ankle and a combination