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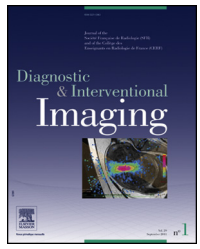

PICTORIAL REVIEW /*Musculoskeletal imaging*

Imaging features of lower limb malformations above the foot

A. Bergère[a], E. Amzallag-Bellenger[a], G. Lefebvre[b], A. Dieux-Coeslier[c], A. Mezel[d], B. Herbaux[d], N. Boutry[a,*]

[a] *Service de Radiopédiatrie, Hôpital Jeanne de Flandre, Université de Lille 2, Centre Hospitalier Universitaire de Lille, avenue Eugène-Avinée, 59037 Lille cedex, France*
[b] *Service de Radiologie et Imagerie Musculosquelettique, Centre de Consultation et d'Imagerie de l'Appareil Locomoteur, Université de Lille 2, Centre Hospitalier Universitaire de Lille, avenue Eugène-Avinée, 59037 Lille cedex, France*
[c] *Service de Génétique Clinique, Hôpital Jeanne de Flandre, Université de Lille 2, Centre Hospitalier Universitaire de Lille, avenue Eugène-Avinée, 59037 Lille cedex, France*
[d] *Clinique de Chirurgie et Orthopédie de l'Enfant, Hôpital Jeanne de Flandre, Université de Lille 2, Centre Hospitalier Universitaire de Lille, avenue Eugène-Avinée, 59037 Lille cedex, France*

KEYWORDS
Congenital morphological abnormalities of the lower limb;
Femur;
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Patella

Abstract Lower limb malformations are generally isolated or sporadic events. However, they are sometimes associated with other anomalies of the bones and/or viscera in patients with constitutional syndromes or disorders of the skeleton. This paper reviews the main imaging features of these abnormalities, which generally exhibit a broad spectrum. This paper focuses on several different bone malformations: proximal focal femoral deficiency, congenital short femur and femoral duplication for the femur, tibial hemimelia (aplasia/hypoplasia of the tibia) and congenital bowing for the tibia, fibular hemimelia (aplasia/hypoplasia) for the fibula, and aplasia, hypoplasia and congenital dislocation for the patella.

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Lower limb malformations (other than the foot) are rare and scarcely described in medical imaging literature. Such malformations are generally isolated events but in some rare instances, they can be associated with other abnormalities of the bones and/or viscera in constitutional syndromes or disorders of the skeleton [1]. Bilateral malformations are generally inherited in an autosomal dominant manner with a varying degree of penetrance

* Corresponding author.
E-mail addresses: nboutry@gmail.com, nathalie.boutry@chru-lille.fr (N. Boutry).

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whereas unilateral malformations are sporadic. In some cases, both the upper and lower leg are involved (e.g., femur abnormality combined with a tibial or fibular malformation of the same limb). Skeletal limb abnormalities can be associated with foot defects (incorrect positioning, malformations and coalitions) or joint abnormalities (hip, knee, and ankle). Soft tissue involvement can also be observed (muscle or tendon abnormalities; more rarely blood vessel and nerve involvement). Lower limb malformations are therefore serious conditions often requiring complicated and prolonged orthopedic treatment, the functional outcome of which is sometimes unsatisfactory.

Limb malformations may result from genetic (mutations) or cytogenetic (abnormal chromosomes) defects, fetal exposure to teratogens (thalidomide, sodium valproate, ethanol, cocaine, X-rays, etc.), viral infection during pregnancy, mechanical forces (amniotic band syndrome) or any combination of the above [2—5].

The majority of lower limb malformations are currently detected during prenatal ultrasonographic examinations. From 26 weeks of amenorrhea, three-dimensional (3D) ultrasonography and low dose fetal computed tomography (CT) [6] can be used for more detailed diagnosis [7]. After birth, X-ray imaging is used predominantly although magnetic resonance (MR) imaging can be very useful to detect and diagnose possible cartilage, soft tissue and articular defects that might be related to the bone malformation.

Formation and development of the lower limbs

Lower limb morphogenesis occurs at an early embryonic stage (between 4 and 6 weeks of life). Lower limb buds appear shortly after upper limb buds (between the 4th and 5th week) as bulges near the lumbar vertebrae [2]. Limb buds are outgrowths of mesenchymal tissue from the mesoderm covered by the ectoderm. In the 6th week, the limb buds lengthen, develop recognizable segments that will become the thigh and lower leg, and the distal ends flatten to form the foot plates. During weeks 7 and 8, interdigital tissue regresses via apoptosis to produce separate toes and the limbs begin to rotate medially.

A number of genes regulate the medial rotation, placement and development of the bud [8]. These include the *TBX4* gene (medial rotation of the lower limb bud), *FGF* (Fibroblastic Growth Factor) genes and their receptors (*FGFR*) (limb bud development), *TP63* (apoptosis) and the *HOX* genes (limb bud type and shape) [8]. Normal limb bud development is three-dimensional and occurs along the proximodistal, anteroposterior (preaxial/postaxial) and dorsoventral axes. The extending limb bud comprises three main signaling centers (Fig. 1) that regulate its development: the apical ectodermal ridge (AER), a structure formed from ectodermal cells at the distal end of the bud, the zone of polarizing activity (ZPA) located in the mesenchyme on the posterior rim of the bud, and the progress zone (PZ) which is adjacent to the AER.

Proximodistal extension of the limb bud is regulated by the AER (and by the interactions that occur between the AER and the ZPA). The morphogen SHH (sonic hedgehog) mediates the polarizing activity of the ZPA, which regulates anteroposterior growth. Finally, dorsoventral development is regulated by the *LMX1B* gene.

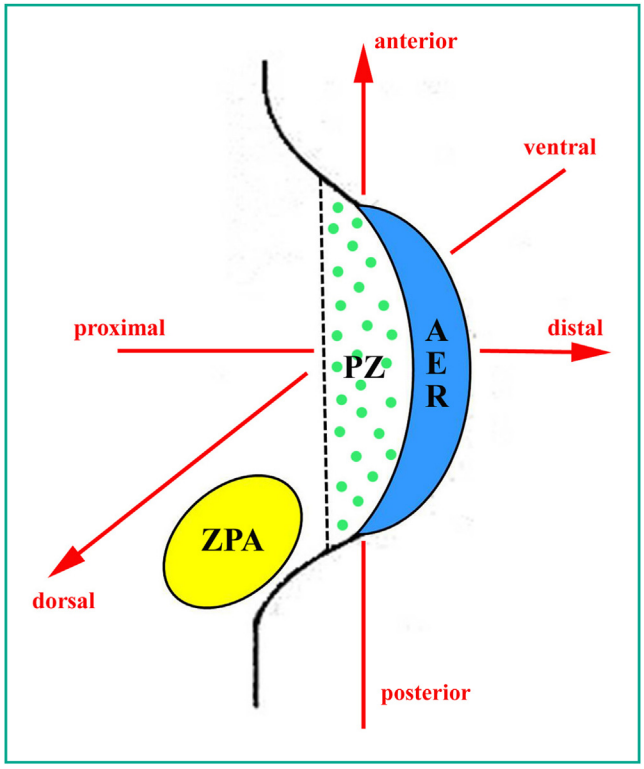

Figure 1. Lower limb bud of a human embryo. AER: apical ectodermic crest; ZPA: zone of polarizing activity; PZ: progression zone.

Congenital defects of the femur

This review focuses on proximal focal femoral deficiency (PFFD), congenital short femur (CSF) and congenital duplication of the femur.

Proximal focal femoral deficiency (PFFD)

This malformation is characterized by a shortened bone due to impaired development of the proximal end of the femur. PFFD is rare (approx. one in 50,000 births) and predominantly unilateral (90% of cases) [9]. The thigh is shortened to a variable degree, the hip and knee joints are flexed [7,8] and the hip may or may not be stable.

X-ray imaging of PFFD patients shows an apparent loss of continuity between the femoral shaft and head/neck. The zone where these structures should converge is made up of fibrous or fibro-cartilaginous tissue that can sometimes ossify at a later stage. Various classifications have been proposed for PFFD. Aitken's classification is the most widely used and includes four classes of PFFD based on severity (A—D) (Fig. 2) [9]:

- class A (least severe form): the femoral head and neck are present, the acetabulum is normal, and a small portion of the proximal femur is missing;

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