Case Report

Association of upper limb meromelia, proximal focal femoral deficiency, fibular hemimelia, and intermetatarsal coalition in a young adult male✩✩∗

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ABSTRACT

Meromelia refers to the partial absence of at least 1 limb and is also referred to as “terminal transverse hemimelia.” It can occur in either isolation or with other congenital malformations. There are very few publications in the literature that report meromelia cases accompanied by other congenital anomalies. Proximal focal femoral deficiency is another rare congenital skeletal abnormality and is characterized by the underdevelopment of the proximal part of the femur and shortening of the entire lower extremity. A case of upper limb meromelia accompanied by proximal focal femoral deficiency and fibular hemimelia in a neonate has previously been reported. However, to our knowledge, the association of upper limb meromelia with intermetatarsal coalition has never been reported to date. Here, we present an adult patient showing an unusual association of multiple rare congenital skeletal abnormalities including meromelia, proximal focal femoral deficiency, fibular hemimelia, and intermetatarsal coalition.

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Introduction

Meromelia refers to the partial absence of at least 1 limb and is also referred to as “terminal transverse hemimelia.” It is a rare congenital skeletal anomaly with a reported prevalence of 1.41 per 100,000 births [1]. Although several etiological mechanisms have been described, the exact cause of the disorder remains unknown. An injury during the early embryonic period, genetic defects in the regulation of limb development, intrauterine amputation as a result of amniotic bands or arterial insufficiency of the extremity have all been addressed as possible causes of meromelia [2]. Most of the reported cases are sporadic. It can occur in either isolation or with other congenital malformations. However, there are very few publications in the literature that report meromelia cases accompanied by other congenital anomalies [2–5].

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Proximal focal femoral deficiency (PFFD) is another rare congenital skeletal abnormality. It is characterized by the underdevelopment of the proximal part of the femur and shortening of the entire lower extremity. The disorder typically presents as a shortened thigh which is flexed, externally rotated, and abducted. The incidence of PFFD is reported to be about 1-2 per 100,000 live births [6]. It usually occurs sporadically and presents unilateral involvement. Up to 60% of the cases are accompanied by other skeletal anomalies, including fibular hemimelia, pes equinovarus, oligodactyly, tibial bowing, absence of the cruciate ligament(s), spinal dysraphism, and microcephaly [7]. However, a case of PFFD in association with intermetatarsal coalition has never been reported to date. Here, we present an adult patient showing an unusual association of multiple rare congenital skeletal abnormalities including upper limb meromelia, PFFD, fibular hemimelia, and intermetatarsal coalition.

Case report

A 20-year-old male patient with bilateral upper extremity and right lower extremity disability applied to the hospital to undergo a mandatory health screening to qualify for disability rights from the state social insurance institution. He had no health complaints other than the skeletal disorders involving his extremities. He is the first of 3 children born to nonconsanguineous healthy parents. His sister and brother are completely healthy. He did not report any type of congenital skeletal anomaly in the extended family. No other significant health problem is present in the history of his childhood. Physical examination revealed that all skeletal elements of both upper extremities were absent except the small proximal humeral segments (Fig. 1a). Length reduction of his right lower extremity was also noted on physical examination (Fig. 1b). He was provided both upper limb prostheses 2 years ago. However, the patient stated that the prostheses were not functional and provided only cosmetic relief. Since he perceived them as a burden, he could not get used to them and did not use them. He reported having had right hip arthrodesis a few years ago to reduce the lateral angulation of the leg. The only functional limb of the patient was his left foot and he was able to eat and write using it.

Anteroposterior (AP) radiograph of both upper extremities showed that all skeletal elements of both arms distal to the proximal rudimentary humeral segments were absent (Fig. 2). Full-length standing AP radiograph of both lower extremities showed that his right lower limb was about 28 cm shorter than its counterpart. No skeletal abnormality was detected in his left lower limb (Fig. 3). Standing AP lumbar spine radiograph revealed pelvic obliquity and scoliosis with the curve convex to the left which were the results of the limb length discrepancy. The fifth lumbar vertebra was sacralized (Fig. 4). AP pelvis radiograph including both femurs demonstrated that the right hemipelvis was smaller than the left side. The right hip joint was fused, and there were several surgical screws at this location due to the previous hip arthrodesis surgery. The right acetabulum, femoral head, and trochanteric structures were absent. The right femur was severely shortened. The

Fig. 1 – Clinical photographs of the patient with upper limb meromelia accompanied by proximal focal femoral deficiency showing his upper (a) and lower (b) extremities.
femur was flexed, externally rotated, and abducted (Fig. 5). AP (a) and lateral-oblique (b) right hip radiographs demonstrated the hip joint fusion, the positions of the screws and severe femoral shortening. The shortening of the right fibula was also evident on the AP radiograph (Fig. 6). AP (a) and lateral-oblique (b) left foot radiographs revealed a coalition between the proximal bases of the fourth and fifth metatarsals (Fig. 7).

Based on highly specific clinical and radiographic findings, the patient was diagnosed as having bilateral upper limb meromelia with right PFFD accompanied by fibular hemimelia and intermetatarsal coalition. Prosthetic use training programs and regular psychotherapy were planned for the rehabilitation of the patient. Besides, he was registered in the social services center and socialization was included in the rehabilitation program. The patient was called for a follow-up visit 6 months later.

**Discussion**

While amelia is the term used to identify the complete absence of a limb/limbs, meromelia refers to the partial absence of a limb/limbs [1]. Since they represent 2 different stages of the same disorder, the term “amelia-meromelia se-
developmental dysfunction case has been previously cited.

"radiographs", 

"Fig. 6 – Anteroposterior (a) and lateral-oblique (b) right hip radiographs clearly demonstrate the hip fusion, the positions of the screws and severe femoral shortening. The right fibula is also shortened (b)."

"Fig. 7 – Anteroposterior (a) and lateral-oblique (b) left foot radiographs showing a coalition between the proximal ends of the fourth and fifth metatarsals."

"sequence" is also used to define the limb reduction of this type [2]. It is shown that 66.9% of the cases with amelia are accompanied by other congenital anomalies [3]. However, publications reporting the associations of meromelia are extremely rare in the current literature [2–5]. The association of meromelia with PFFD and fibular hemimelia has previously been shown [4]. And a case of meromelia with oligodactyly and brachymesophalangy of the foot has also been reported [2]. However, to our knowledge, this is the first case report of meromelia accompanied by intermetatarsal coalition.

PFFD is characterized by the shortening, deformity, and dysfunction of the extremity which are the results of the developmental failure of the subtrochanteric femoral segment. The disorder presents a wide clinical spectrum ranging from mild cases with minimal femoral shortening and normal hip development to the most severe cases with complete femoral absence. Currently, the most widely used PFFD classification is the one proposed by Aitken and modified by Amstutz [6]. It has been reported that other congenital abnormalities, especially fibular hemimelia, accompany PFFD in 30%-60% of all cases [7,8]. Fibular hemimelia and intermetatarsal coalition were also present in our patient.

Congenital coalitions of the foot are relatively uncommon abnormalities and mostly develop in the hindfoot and midfoot [9]. Coalitions between the forefoot bones, especially between metatarsals, are extremely rare with only a few reported cases in the current literature [9–16]. While most of the reported cases were in isolation, cases of the intermetatarsal coalition as a component of Pfeiffer syndrome [17,18], have also been reported. Intermetatarsal coalition mostly develops between the base of the fourth and fifth metatarsals and, less commonly, between the first and second metatarsals. The disorder may be asymptomatic like it was in our patient. However, it may cause metatarsalgia and disability in patients with impaired metatarsal mobility [10].

We presented an unfortunate young man who was suffering from a combination of 2 major skeletal abnormalities accompanied by relatively minor disorders. Causing not only a significant physical disability but also a serious cosmetic discomfort, such severe skeletal abnormalities require comprehensive treatment planning. Supplementation of a prosthesis or orthosis is indicated in the rehabilitation of children with limb deficiencies [2]. Our patient was neglected and missed the correct times for appropriate treatment interventions. Only 2 years ago, upper limb prostheses were provided and he could not cooperate with the use of devices. For the treatment of right PFFD, surgical correction was performed only a few years ago and the lower extremity was not supported by a prosthesis. The patient’s ambulatory skills were extremely limited and his personal care was being provided by others.

In addition to surgical intervention(s) and prosthesis instrumentation, other needs, including the education and psychological status of the child, should be carefully considered in the management of patients with limb defect(s). Specialized and interdisciplinary teamwork is essential to comprehensively address all these critical issues and to be able to intervene at the appropriate time. The ideal team to undertake this task is a large team including a physician, orthopedist, prosthetist, physical therapist, occupational therapist, psychologist, and especially the mother of the child [2].

Informed consent

Informed consent for publication was obtained from the patient.

Supplementary material

REFERENCES


