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CASE REPORT

Ulnar hemimelia: a report of four cases

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Abstract

Ulnar hemimelia is a very rare skeletal abnormality characterized by the total or partial absence of the ulna. It is reported to occur in approximately 1 per 150,000 live births. Some shortening of the forearm, radial bowing, and tendency of the hand to drift to the ulnar side of the wrist usually accompany ulnar hemimelia. Other skeletal anomalies such as humeroradial synostosis, radial head dislocation, carpal or metacarpal coalition, and digital abnormalities may also be seen in cases of ulnar hemimelia. The patients may be asymptomatic in the presence of an isolated mild ulnar deficiency. On the other hand, cases of prominent ulnar deficiency accompanied by complex upper limb abnormalities leading to severe disability may also be observed. We herein present four patients with varying degrees of ulnar hemimelia. Our first case had an isolated ulnar hemimelia, whereas the other three had additional upper limb abnormalities of different types.

Keywords Congenital · Hemimelia · Limb anomaly · Radiography

Introduction

Hemimelia is the term used to describe the congenital skeletal abnormality characterized by the absence or underdevelopment of one side of the distal half of a limb. Occurring in approximately 1 per 150,000 live births, ulnar hemimelia is a very rare form where the ulna is totally or partially absent [1]. The reason for its rarity compared with other skeletal anomalies is speculated to be its relatively early development in embryonic life. The critical embryonic period in the development of ulnar deficiency (24–26 days) is earlier than those of other skeletal anomalies and it corresponds to a period of high mortality [2, 3]. Ulnar hemimelia is more common in men, with a male to female ratio of 3:2. It is reported to be unilateral in about 70% of cases, and is mostly right-sided and incomplete. Some shortening of the forearm, radial bowing and tendency of the hand to drift to the ulnar side of the wrist usually accompany ulnar hemimelia. Other skeletal anomalies such as humeroradial synostosis, radial head dislocation, carpal or metacarpal coalition, and digital abnormalities may also be seen in cases of ulnar hemimelia. Different from the radial deficiencies, ulnar deficiency is mostly nonsyndromic and is most commonly seen in skeletal dysplasias. The radius is typically longer than the ulna in patients with achondroplasia, and in patients with mucopolysaccharidosis, the distal ulna and radius can be hypoplastic and may slope toward each other [4, 5]. Ulnar hemimelia may also present with Poland syndrome, Goltz–Gorlin syndrome, Cornelia De Lange syndrome, or femur fibula ulna syndrome [1, 6].

Patients with ulnar hemimelia may be asymptomatic in the presence of isolated mild ulnar deficiency. On the other hand, cases of prominent ulnar deficiency accompanied by complex upper limb abnormalities leading to severe disability may also be observed. Herein, we present four patients with varying degrees of ulnar hemimelia. Our first case had an isolated ulnar hemimelia, whereas the other three had additional upper limb abnormalities of different types.

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Case reports

Case 1

A 34-year-old woman presented with the complaint of a mild intermittent pain in her left elbow for the last few months.

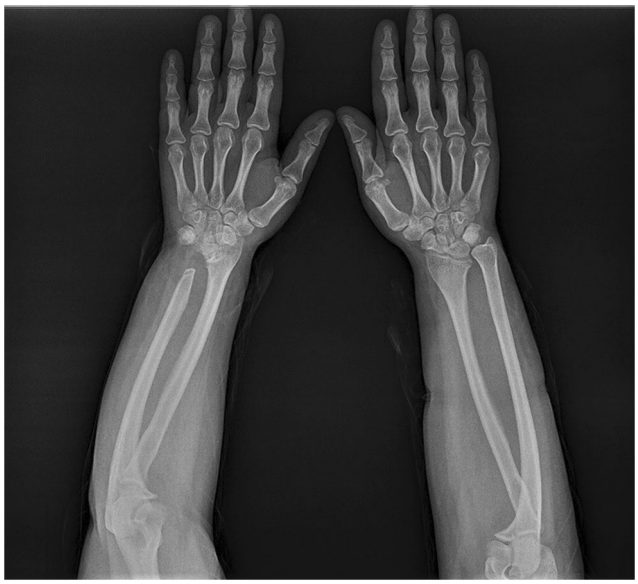

Fig. 1 Anteroposterior radiograph of both forearms including the hands and the elbows. The right forearm, right elbow joint, and both hands are radiographically normal. However, the distal portion of the left ulna is partially absent

There was no history of trauma to her left arm during her childhood or later years. On physical examination, no loss of function was recorded in her left upper limb.

Anteroposterior (AP) radiograph of both forearms including the hands and the elbows were obtained. The right forearm, elbow joint and hand were radiographically normal (Fig. 1). AP and lateral (L) radiographs of the left forearm revealed partial absence of the distal portion of the ulna (Fig. 2a, b). There was some periarticular sclerosis consistent with mild osteoarthritis in the left humeroulnar joint (Fig. 2c). No anomaly was noted in the left hand.

Case 2

A 20-year-old man presented with pain and restriction of movement of his left elbow. He stated that he had been suffering from elbow pain for the last few years, but the restriction of elbow motion had begun only a few months ago. There was no trauma or any significant feature in his childhood or later history. On physical examination, the third digit of the left hand was short, and the left elbow extension was painful (Fig. 3a).

Anteroposterior and lateral radiographs of both forearms (including hands) were obtained. The right forearm and hand were radiographically normal (Fig. 3b). The radiographs of the left forearm revealed the partial absence of the distal portion of the ulna and radial bowing. Camptodactyly in the third digit, accompanied by the bowing of the third metacarpal, was evident in the left hand radiographs (Fig. 4a, b). There were intense periarticular sclerosis and prominent narrowing in the left humeroulnar joint consistent with severe osteoarthritis (Fig. 4c, d).

Case 3

A 26-year-old woman presented with a limited range of motion of her elbows and cosmetic disturbance concerning her upper limbs. She stated that her both arms had been curved, but had functioned well since childhood. She had been offered surgery when she was about 10; however, she could not

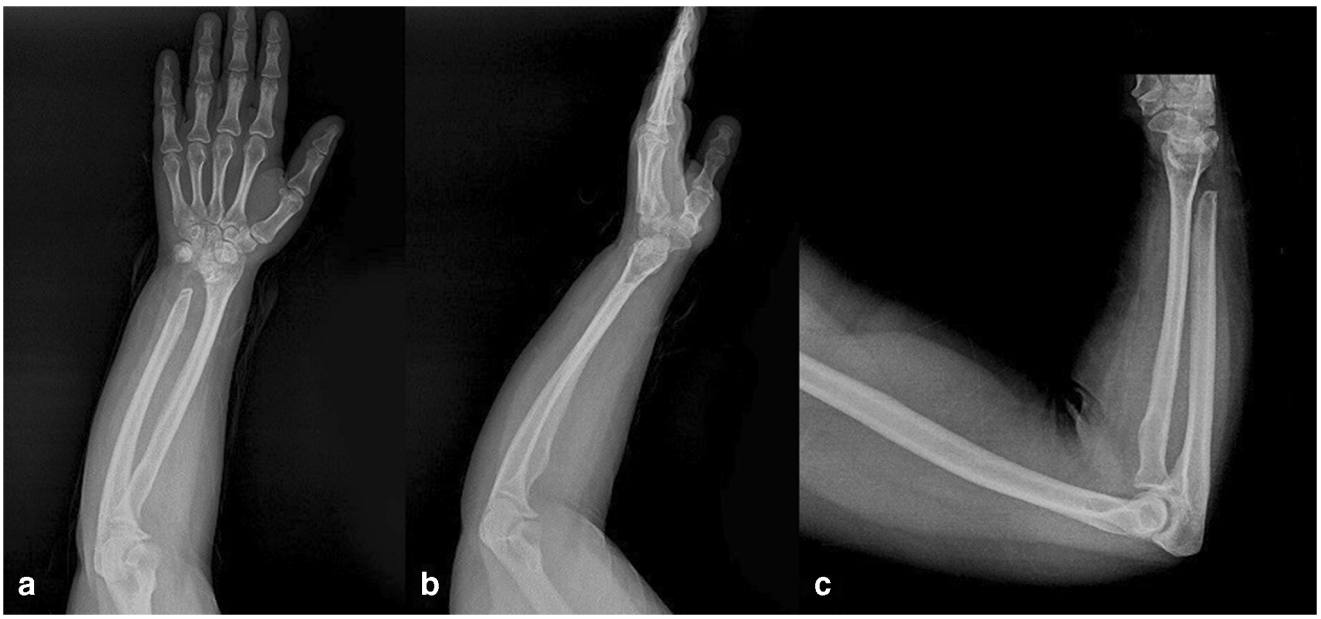

Fig. 2 **a** Anteroposterior and **b** lateral radiographs of the left forearm demonstrate partial absence of the distal portion of the ulna. **c** Lateral radiograph of the left elbow joint depicts some periarticular sclerosis consistent with mild osteoarthritis in the humeroulnar joint

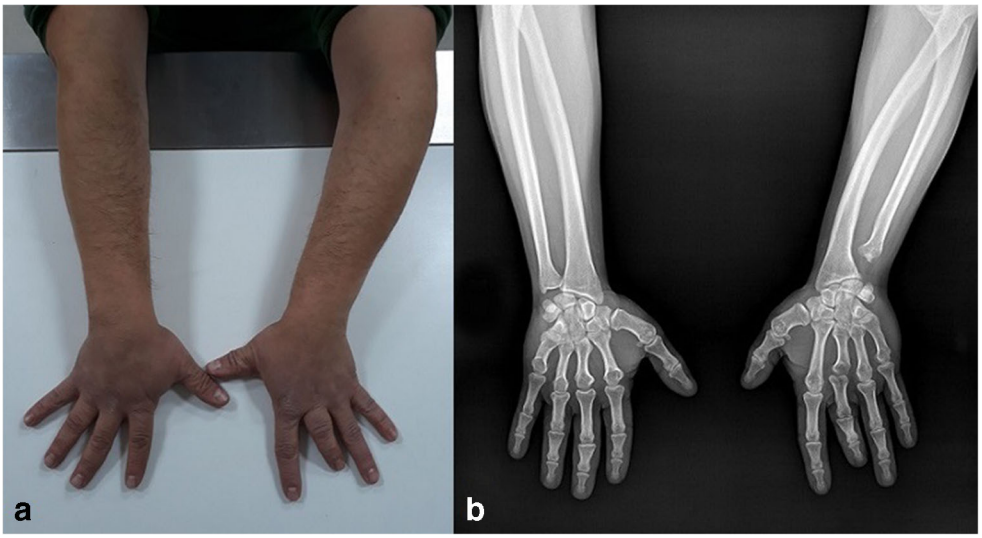

Fig. 3 **a** Clinical photograph and **b** anteroposterior radiograph of both forearms, including both hands, show that the right forearm and hand are normal, whereas the third digit of the left hand is short. Partial absence of the distal portion of the left ulna, and left radial bowing is evident on the radiograph (**b**)

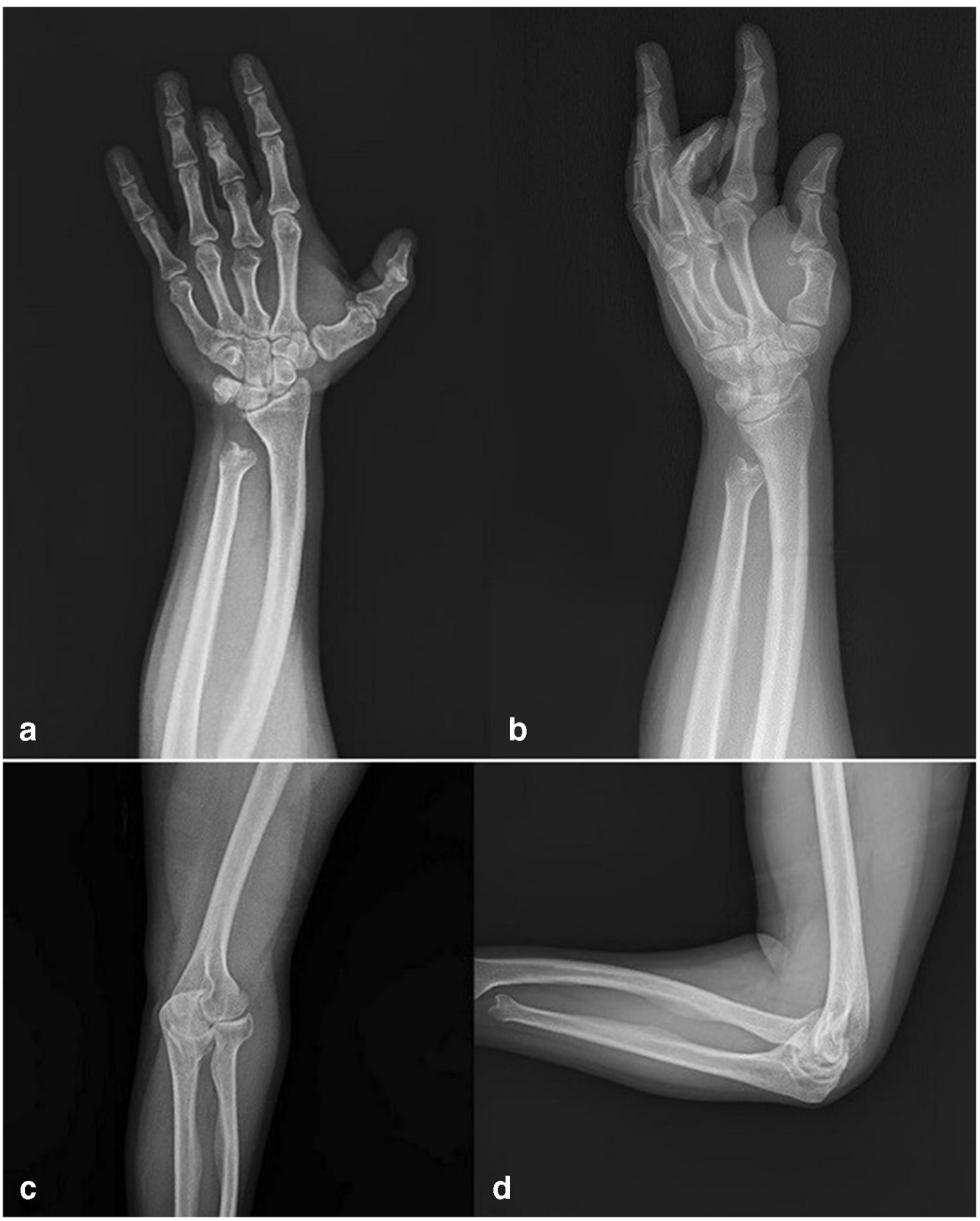

Fig. 4 **a** Anteroposterior and **b** lateral radiographs of the left forearm depict the partial absence of the distal portion of the ulna, and radial bowing. Camptodactyly in the third digit, accompanied by the bowing of the third metacarpal, is evident in the lateral radiograph (**b**). **c** Anteroposterior and **d** lateral radiographs of the left elbow joint show intense periarticular sclerosis and prominent narrowing in the humeroradial joint consistent with severe osteoarthritis

undergo surgery because of financial difficulties. She had no systemic complaints other than the two upper limb deformities. On physical examination, the pronation and supination of the radioulnar joint and elbow movements were restricted on both sides. No sensory deficit was noted (Fig. 5).

Anterior radiographs of both forearms, including both elbows, along with AP radiographs of both hands, were obtained. The distal half of both ulnar bones were absent, and there was fixed flexion of about 45° of both elbow joints. Dislocation of the radial heads, bilateral radial bowing, and mild ulnar-sided drift of the hands were demonstrated (Fig. 6a, b). Fixed flexion of varying degrees of the digits was observed on the radiographs of both hands (Fig. 6c, d).

Case 4

A 24-year-old man presented with severe pain and restriction of movement of his left shoulder. Both upper limbs were short, and bilateral hand oligodactyly was present. He stated that he had never undergone surgical or nonsurgical treatment for the deformities involving his upper extremities. He had no systemic complaints other than musculoskeletal deformities and shoulder pain. Physical examination revealed fixed extension of both elbow joints, and left shoulder instability.

Anteroposterior and lateral radiographs of both forearms, including hands and both elbows, were obtained, along with the AP radiographs of both shoulders. Bilateral ulnar hypoplasia accompanied by bilateral shortening of the limb, humeroradial synostosis, radial bowing, massive carpal coalition, and tridactyly were depicted. The limb shortening was more prominent on the left side (Fig. 7a, b). In both hands, the proximal phalanges of the third digits were bifid, and the medial and distal phalanges of the third digits were duplicated. There was fixed flexion of varying degrees of the digits of both hands (Fig. 7c, d). AP radiograph of the right shoulder was normal. AP radiograph of the left shoulder revealed glenohumeral dysplasia of the shoulder with a dysplastic and hypoplastic glenoid, a small humeral head, a small scapula and arm, a blunted coracoid process, and muscular atrophy in the arm. Minor rotoscoliosis with the curve convex to the right, L4 butterfly vertebra, and spinal dysraphism at the T12 level were also noted (Fig. 8).

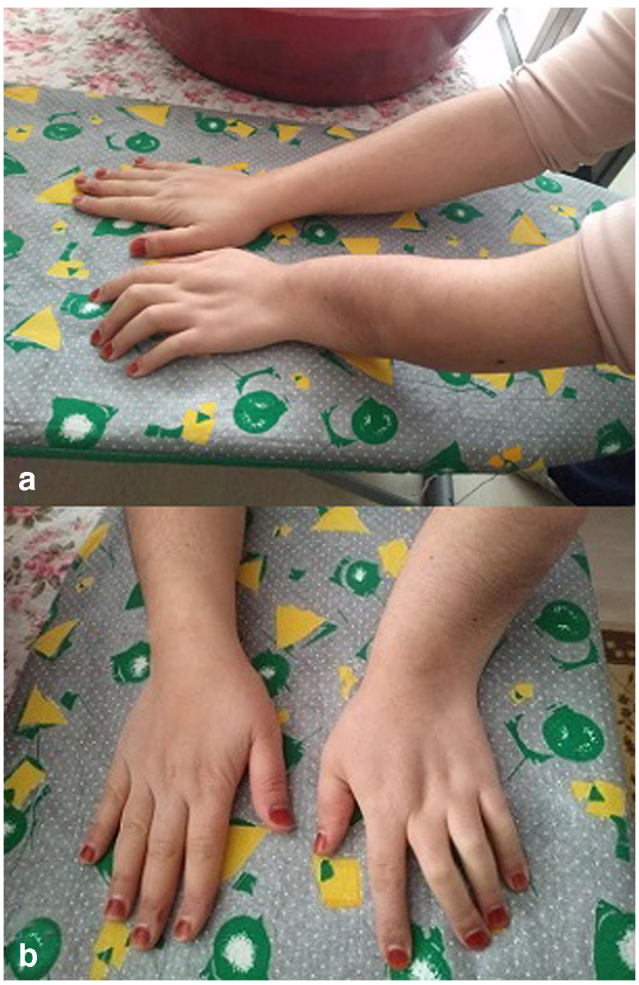

Fig. 5 Clinical photographs of both **a** forearms and **b** hands show that both arms are curved inwards, and there is a mild ulnar-sided drift of the left hand. There are also varying degrees of fixed flexion of the digits of the left hand

Discussion

We presented four patients with ulnar hemimelia, two of whom were admitted with relatively mild clinical complaints of pain and restricted motion of a single elbow joint, whereas the other two were suffering from severe disability of both upper limbs and serious cosmetic discomfort. The severity of the ulnar deficiency and the presence or absence of the accompanying skeletal anomalies determine the time of presentation and the clinical course of ulnar hemimelia. The two main complaints of the patients are disability of the limb and cosmetic disturbance. The limitation of the extension of the elbow joint to approximately 90° is a frequent finding among patients with ulnar hemimelia. In severe cases, fixed flexion of the joint up to 160° may be present [1]. In our third case, there was fixed flexion of about 45° of both elbow joints, whereas our fourth case presented bilateral humeroradial synostosis with bilateral fixed extension of the elbow joints. Fixed flexion or extension of the elbow joint is reported to occur in cases with accompanying humeroradial synostosis. Congenital bilateral humeroradial synostosis is a rare skeletal disorder associated with ulnar hypoplasia and is classified according to the position of the forearm. Class I is sporadic and comprises patients with fixed extension, whereas class II is familial and comprises patients with fixed flexion. Class II is reported to be associated with multiple systemic anomalies [7, 8]. Humeroradial synostosis of our patient (case 4) was of class

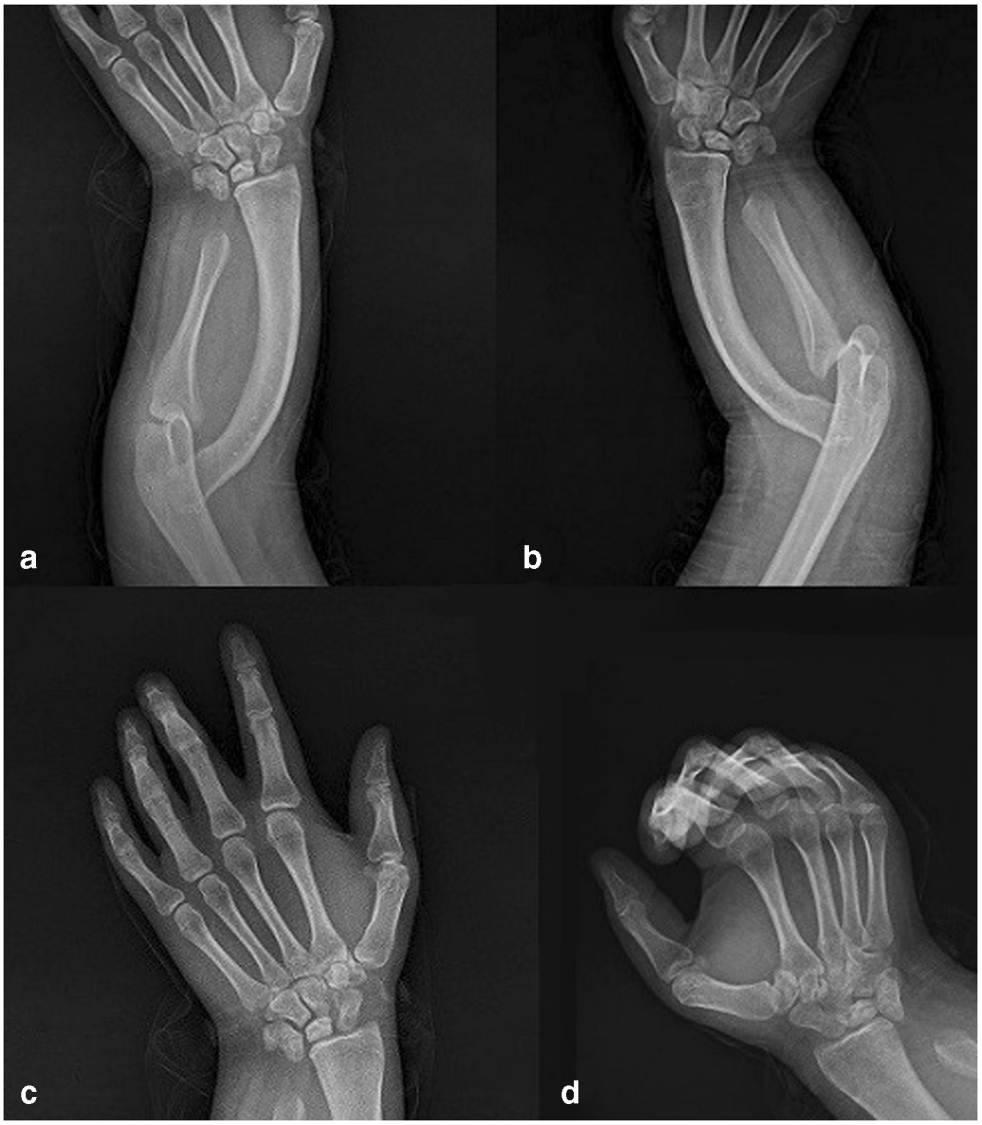

Fig. 6 Anteroposterior radiographs of **a** the right and **b** the left forearm, and anteroposterior radiographs of **c** the right and **d** the left hand. The distal halves of both ulnar bones are absent, and there is fixed flexion of about 45° of both elbow joints. Dislocation of both radial heads, bilateral radial bowing, and mild ulnar-sided drift of the hands are also evident (**a**, **b**). Fixed flexion of the digits of varying degrees is seen (**c**, **d**)

I, and caused severe disability of both upper limbs of this young man. However, the reason for bringing the patient to the doctor was the recurrent pain in his left shoulder. Shoulder radiography revealed glenohumeral dysplasia, implying the presence of shoulder instability. To our knowledge, this is the first case reported to have glenohumeral dysplasia associated with ulnar hemimelia. Tridactyly in both hands of this unfortunate patient increased the severity of his disability.

Ulnar hemimelia may be associated with complex carpal, metacarpal, and digital abnormalities. Carpal abnormalities including aplasia, hypoplasia and fusion may accompany ulnar deficiency. The triquetrum and the capitate are commonly absent in these patients [9]. In addition, the absence of postaxial (ulnar-sided) metacarpal and digital bones are frequent findings in patients with ulnar hemimelia. The most common hand anomaly associated with the disorder is three-fingered hand (tridactyly), closely followed by mono-digital hand [2, 9]. Our fourth case presented all these mentioned features. The number of the deficient digits and the presence or absence of additional digital abnormalities such as syndactyly and camptodactyly (fixed flexion deformity of the interphalangeal joints) closely affect the functionality of the hand [2, 6]. Camptodactyly was present in our second case, and fortunately did not cause serious disability, as the neighboring digits functioned well. In syndromic cases of ulnar hemimelia, the clinical course and the prognosis are reported to depend on the severity of the syndrome [10]. In none of the patients in our series was ulnar hemimelia associated with a syndrome.

The disability caused by ulnar hemimelia and associated anomalies deteriorates in adulthood as disuse atrophy and osteoarthritis are added to the clinical picture. We detected severe osteoarthritis in the humeroradial joint of our second case who was only 20 years old. Early diagnosis and applying an appropriate treatment of ulnar hemimelia is essential, and the diagnosis can be based solely on radiography. Management of the disorder is challenging, and should be highly

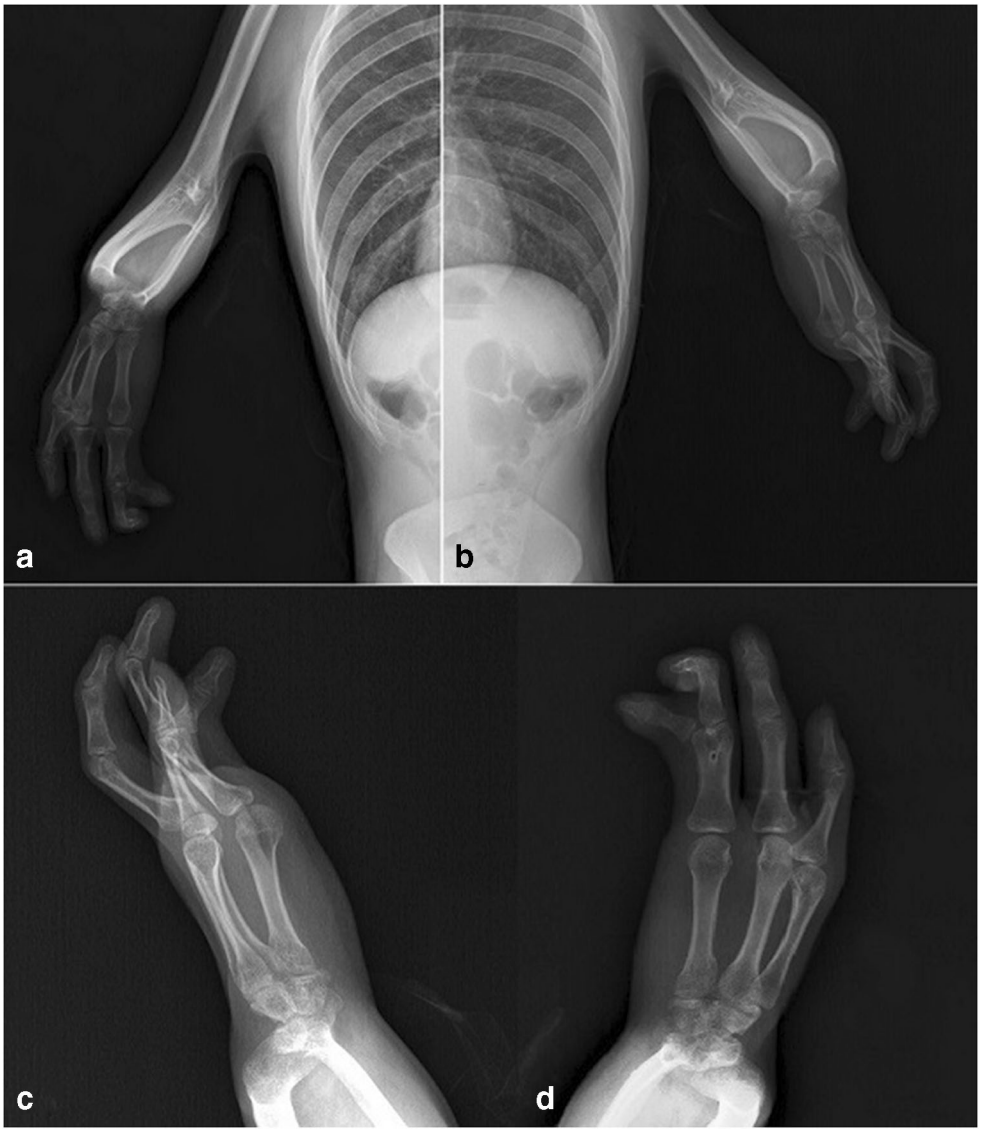

Fig. 7 Anteroposterior radiographs of **a** the right and **b** the left forearm, and anteroposterior radiographs of **c** the right and **d** the left hand. Bilateral ulnar hypoplasia accompanied by bilateral shortening of the limb, humeroradial synostosis, radial bowing, massive carpal coalition, and tridactyly are depicted. Note the limb shortening is more prominent on the left side (**a**, **b**). In both hands, the proximal phalanges of the third digits are bifid, and the medial and distal phalanges of the third digits are duplicated. There is fixed flexion of the digits of both hands of varying degrees (**c**, **d**)

individualized. The most important factors determining the treatment approach are the severity and complexity of the abnormality, the age of the patient, and the uni-/bilaterality of the disorder. Continuous physiotherapy and training should

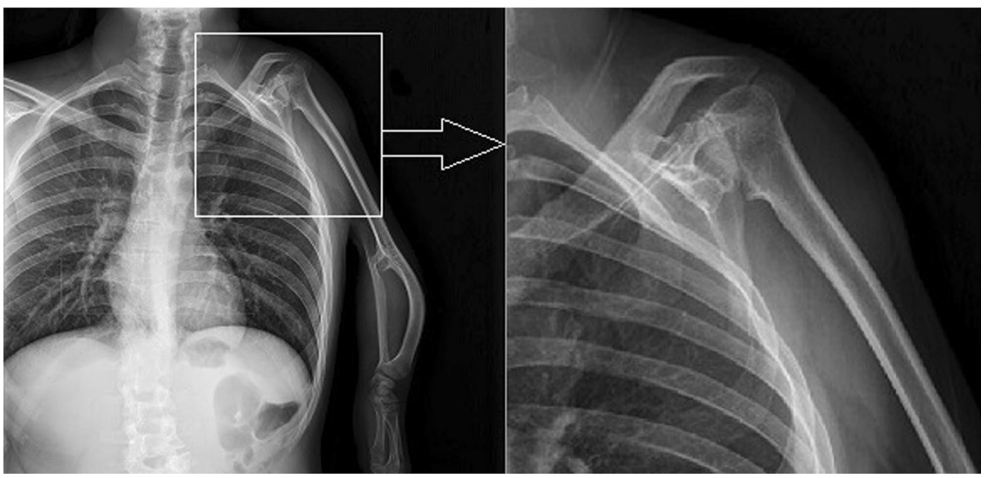

Fig. 8 Anteroposterior radiograph of the left shoulder joint demonstrates glenohumeral dysplasia of the shoulder with a dysplastic and hypoplastic glenoid, a small femoral head, a small scapula and arm, a blunted coracoid process, and muscular atrophy in the arm. Minor rotoscoliosis with the curve convex to the right, L4 butterfly vertebra, and spinal dysraphism at the T12 level are also evident

be given from infancy to avoid disuse atrophy and to obtain maximum functioning of the limb. For the improvement of function in patients with varying degrees of ulnar hemimelia, several nonsurgical management approaches, including elbow prothesis, are available from puberty [1, 2]. In our first and second cases, regular and continuous physiotherapy was applied. In both patients, successful results were obtained in terms of both pain relief and improvement of function. In cases with bilateral involvement and significant reduction in the range of motion, surgical intervention is indicated. Different surgical approaches including Z-plasty, elbow disarticulation, and humeral derotational osteotomy may be performed to improve the range of motion of the elbow joint [1, 2]. A humeral rotational osteotomy was planned for our third case to achieve cosmetic and functional improvement. For our fourth case, glenoid augmentation (bone graft) surgery was performed to create a blocking effect for the patient's instability; however, restriction of movement and pain of the shoulder joint developed early after the surgical intervention.

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