Case Report

An unusual phenotype of radial longitudinal deficiency (radial hemimelia) presenting in a young adult male

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A B S T R A C T

Radial longitudinal deficiency is a rare skeletal anomaly characterized by a defect in the development of structures that form the radial half of the forearm. The disorder is associated with a large spectrum of preaxial abnormalities. It is shown that the thumb and preaxial carpal bones are almost always hypoplastic or absent in almost all types of radial longitudinal deficiency. Congenital dislocation of the dysplastic radial head may accompany this rare deformity. Herein, we present a 20-year-old male patient with radial longitudinal deficiency who had a markedly hypoplastic radius but had a thumb and carpal bones with normal size, shape, and joint relations. Further, the right radial longitudinal deficiency of our patient was unusually accompanied by left congenital radial head dislocation. Our case shows that, although rare, radial longitudinal deficiency can present without any carpal and thumb abnormalities. And the current case also shows that a contralateral sided congenital radial head dislocation may accompany radial longitudinal deficiency.

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Introduction

Radial longitudinal deficiency (RLD) is characterized by a defect in the development of structures that form the radial half of the forearm. The disorder is also expressed in various terms such as radial hemimelia, radial dysplasia, radial meromelia, or radial club hand. However, given the frequently associated abnormalities of the upper limb, the term RLD may be more revealing to describe the disorder [1]. It is a rare anomaly with an incidence ranging between 1/30,000 and 1/100,000 live births. Bilateral involvement is present in about half of the cases. While RLD is isolated in approximately one-third of the reported cases, the remaining 67% are syndromic or accompanied by systemic or skeletal abnormalities [2]. The most common skeletal abnormalities associated with RLD are scoliosis,
Fig. 1 – Clinical photograph of the patient with RLD in the right side. There is also a slight fixed flexion of the left elbow joint.

humeral hypoplasia, proximal radioulnar synostosis (PRUS), congenital radial head dislocation (CRHD), and camptodactyly.

RLD is associated with a large spectrum of preaxial abnormalities ranging from mild deficiency of radial-sided fingers to complex deformities involving the preaxial elbow, forearm, wrist, and digital joints. It is shown that the thumb and preaxial carpal bones are hypoplastic or absent in almost all types of RLD [3–6]. Furthermore, carpal bone deficiency was accused of being partially responsible for the radially deviated hand, a feature of RLD cases [3]. Herein, we present an unusual case of RLD who had a markedly hypoplastic radius but had a thumb and carpal bones with normal size, shape, and joint relations. In addition, the right RLD of our case was unusually accompanied by left CRHD.

Case report

A 20-year-old male with right upper limb disability admitted to our hospital for mandatory health screening before military service. He had no health complaints other than the skeletal disorder involving his right upper limb. In detailed questioning, he stated that he had a slight pain and restriction of movement in his left elbow. He is the last of 6 children born to nonconsanguineous healthy parents. All of his brothers and sisters are completely healthy. There is no family history of any kind of congenital skeletal abnormalities in the extended family. No other significant health problem is present in the history of his childhood. During childhood, he was offered surgical treatment for several times. However, because of the financial difficulties and his family’s inability to show sufficient care, he had never undertaken any kind of treatment. He was experiencing serious trouble in performing daily activities using his right hand. Physical examination of the right upper extremity revealed fixed extension of the elbow, and fixed radial deviation of the wrist joints. Of the left elbow joint, movements were restricted and extension was painful (Figs. 1 and 2).

Anteroposterior (AP) radiograph of the right upper extremity demonstrated the absence of the proximal third of the radius, an ulnar curve with the convexity toward the ulnar side, and radial deviation of the wrist joint (Fig. 3). All the carpal, metacarpal, and phalangeal bones were present and were of normal size and shape. Distal radial epiphysis was well developed. AP and lateral (L) radiographs of the right elbow depicted the fixed flexion of the joint as well as the obliteration of the joint space distance (Fig. 4). AP and L radiographs of the left elbow showed fixed flexion of about 30” of the joint. Radial head was posteriorly dislocated. There was some periarticular sclerosis consistent with mild osteoarthritis in the ulnohumeral joint (Fig. 5A and 5B). AP left-hand radiograph was normal (Fig. 5C).

Based on the characteristic radiographic findings, the patient was diagnosed as having right RLD and left CRHD. He was then referred to the Department of Orthopedics for the planning and implementation of appropriate treatment.
Discussion

Characteristic components of RLD phenotype can be listed as follows: a hypoplastic radius, a curved ulna, a short forearm, a radially deviated wrist, deficiency of the preaxial carpal bones, and varying degrees of hypoplasia or absence of the thumb [1]. The radiographic classification system includes six types [3,7]: type N is a normal radius and carpal bones with thumb hypoplasia; type O is a normal radius with radial-sided carpal deficiency; type I is a short radius characterized by a distal radial physi displacement > 2 mm proximal to the distal ulnar physis; type II is a very short radius with underdevelopment of both proximal and distal epiphyses; type III is partial absence of the radius (defect can involve proximal, middle, or distal third, but usually distal); type IV is the complete absence of the radius. The case we currently present appears to be consistent with type III. Many investigators have reported that preaxial carpal bone deficiency and thumb abnormalities are almost always present in all types of RLD except for the type N in which the carpal bones are normal [3–6]. Further, Flatt et al described the deformity as “a profoundly abnormal hand joined to a poor limb by a bad wrist” [8]. However, contrary to these definitions, our case showed no abnormality in the wrist or thumb. The radial deviation of the wrist occurs as the result of the lack of mechanical support of the wrist due to the underdevelopment of both distal radial epiphysis and radial-sided carpal bones [3]. However, as a striking finding, our case showed a marked radial deviation although both the distal radial epiphysis and radial-sided carpal bones were all well developed. The deviation was caused by the sharp angulation of the distal radius itself. Therefore, our case shows that, although rare, RLD can present without any carpal and thumb abnormalities. And the
current case also shows that a prominent radial deviation of the wrist can occur even in the presence of well-developed both distal radial epiphysis and radial-sided carpal bones.

Although rare, CRHD is the most common congenital anomaly of the elbow. It usually presents with a decrease in the range of motion of the elbow joint or elbow pain. Dislocation is toward posterior in 65% of the patients. CRHD can occur in either isolation or more commonly in association with other disorders or syndromes [9]. It has been reported that CRHD may develop in case the length of the radius is inadequate [10]. In their retrospective study carried out in a large population of patients with RLD, James et al recorded CRHD or PRUS in 44% of type 1 and 11% of type 0 cases [3]. They noted neither of these anomalies in the cases of type N, 2, or 3.

Given the results of this study, our case with type 3 RLD is not a strong candidate for an accompanying CRHD. However, interestingly, he had CRHD in contralateral side. Our case shows that, even if it is seemingly normal, the contralateral upper extremity of the patients with RLD should be carefully examined in order not to miss a minor abnormality such as CRHD or PRUS.

It has been reported that, over time, an uncorrected RLD leads to the loss of hand function and cause the hand to become a terminal device used only to capture objects between the arm and forearm. Therefore, the treatment of the disorder should be started immediately after birth. Stretching and splinting are the best conservative treatment choices to avoid the wrist tightness. The forearm deformity of children with RLD is corrected by surgically positioning the hand and carpal bone(s) on top of the ulna with the use of centralization or radialization methods [2]. However, in neglected or recurrent cases among adolescents, these methods will no longer be useful and salvage ulnocarpal arthrodesis is indicated [1]. In CRHD cases, observation is a widely accepted treatment approach since childhood symptoms are usually mild. Surgery is indicated only in cases with pain, functional impairment, or serious cosmetic discomfort. Various surgical methods including resection, rotation osteotomy, ulnar osteotomy, and reconstruction of the annular ligament can be performed for the treatment of CRHD [9]. Unfortunately, our patient missed all the suitable age periods for the effective treatment of both deformities and became an adult without receiving any conservative or surgical treatment to date.

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