Ulnar Longitudinal Deficiency

10

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Introduction

Ulnar longitudinal deficiency (ULD) is a rare condition that usually affects the entire upper limb, including the elbow, forearm, and hand. It has been reported to occur in 1:25,000 live births and ULD is most commonly unilateral [1, 2]. It is a sporadic, non-inherited condition, but can be associated with other musculoskeletal anomalies, such as proximal femoral focal deficiency, fibular and tibial deficiency, scoliosis, and finger differences [1, 2].

Embryology

To better understand the clinical appearance and variation in the spectrum of ULD, one must first review the development of the upper limb. Starting around days 26–52 after fertilization, the limb bud develops around three axes: proximal-distal, dorsal-ventral, and radial-ulnar [1, 3–5]. Each axis has its own signaling center:

1. Apical ectodermal ridge (AER) coordinates the proximal-distal outgrowth
2. Zone of polarizing activity (ZPA) controls radial-ulnar asymmetry
3. Progress zone (PZ) for dorsal-ventral differentiation [3, 6]

Integral to these specialized zones are several signaling molecules. They include fibroblast growth factors, sonic hedgehog, and bone morphogenic proteins. These molecules affect each other through feedback loops [3]. In regard to ULD, sonic hedgehog is responsible for development of ulnar-sided forearm structures as well as the four ulnar-sided digits [1]. The thumb abnormalities occasionally seen in ulnar dysplasia can be explained by the sonic hedgehog-fibroblast growth factor feedback loop [6].

Classification and Clinical Picture

The spectrum of clinical presentation for ULD is quite variable. The entire upper limb is involved in the majority of cases with classification schemes focusing on the elbow/forearm abnormalities [7–11], hand [12], and more specifically the thumb and first web space [13].

The commonly used Bayne classification [7] describes the progression of deficiency noted at the elbow and forearm. Its original description had four types and was later modified by Havenhill et al. [8]. This modified classification of ULD is as follows:

1. Normal length ulna with ulnar-sided hand anomalies
2. Hypoplasia of the ulna (presence of distal and proximal ulnar epiphysis)
3. Partial aplasia of the ulna (absence of the distal or middle one third of the ulna)
4. Total aplasia of the ulna (complete absence of the ulna)
5. Complete absence of the ulna with radiohumeral synostosis (fusion of the radius to the humerus)

Goldfarb et al. [14] proposed a type V ulnar longitudinal dysplasia that would incorporate cases of severe radiohumeral synostosis with humeral bifurcation or a large medial epicondyle. Given the rarity of the disease along with the variable presentation, Buck-Gramcko [15] stated that the pathological findings in ulnar deficiency are so different in the involvement and distribution in the arm, it is impossible to divide them into a classification system. Although Bayne and others describe the elbow and forearm abnormalities, treatment has really been focused more on the hand and digits. As such, Cole and Manske [13] presented a classification

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system based upon the characteristics of the thumb and first web:

1. Normal first web space and thumb
2. Mild first web and thumb deficiency
3. Moderate to severe first web and thumb deficiency; potential loss of opposition; malrotation of the thumb into the plane of the other digits; thumb–index syndactyly; absent extrinsic tendon function
4. Absent thumb

The authors of this classification scheme opine that it is the complexity of the radial-sided problems that require the majority of surgical procedures, and as such this classification will focus the surgeon's attention on those deficiencies that are most important for the restoration of function. They concluded that ULD be classified by an elbow/forearm system and supplemented by the hand classification [13].

Associated Anomalies

Unlike radial longitudinal deficiency, patients with ULD rarely have heart or hematopoietic anomalies. However, these patients may have associated musculoskeletal anomalies such as proximal femoral focal deficiency, additional hip pathology (coxa vara), tibial or fibular ray deficiency, phocomelia, scoliosis, clubfeet, and spina bifida [1, 16].

Upper Arm and Shoulder

With regard to the proximal humerus and shoulder region, hypoplasia can occur. Despite the abnormality, most patients do not have restricted motion [15].

Elbow

There is quite a bit of variation with regard to clinical presentation. The elbow may be stable, unstable, or fused. The articular surfaces can range from normal to hypoplastic to severely deformed. Congenital dislocation of the radial head can be present and subsequent deformity to the distal end of the humerus present [15]. El Hassan et al. [17] reported that 12 % of ULD had a radiohumeral synostosis. In their series, the elbows were fixed in 20–90° of flexion, with no elbows in full extension [17]. Others have described the elbow fixed in full extension [11] or severe flexion and rotation of the elbow so that the hand is positioned behind the child and away from the opposite, uninvolved hand, the so-called hand on flank deformity [18].

Forearm

Buck-Gramcko [15] reported that the different types of ulna defect show no correlation to the severity of the involvement of other parts of the arm and can be combined with all variations of hand and elbow anomalies. The majority of patients with ULD will have a shorter forearm than normal (Fig. 10.1). Havenhill et al. [8] described a variation of patients with a normal forearm but deficiencies isolated to the ulnar side of the hand—Type 0. In ULD, ulnar hypoplasia is most common (60 %) with partial absence of the ulna reported 22.5 % and complete absence in 18 % of patients [15]. Some patients with ULD will have a fibrocartilaginous mass, possibly representing the anlage of the absent portion of the ulna [16]. This is commonly seen in Bayne Types II and IV and may be the cause for radial bowing and wrist deviation, although this point has been debated [16–22].

Wrist

Angulation of the wrist can occur, but is typically not as severe as seen in radial longitudinal deficiency. (Fig. 10.1c). El Hassan et al. [17] reported that the wrist was positioned in neutral in 71 % of patients, with the remaining wrists resting in 5–40° of ulnar deviation. Those patients with the wrist in neutral position had essentially normal wrist range of motion. However, when the wrist was in ulnar deviation, limitations were documented with regard to radial deviation, wrist flexion, and extension [17]. Controversy over the role of the ulnar anlage and the amount of wrist deviation exists [15–17]. Carpal bones can be absent in correlation with missing digital rays, and synostoses can occur in 30–40 % of cases [15].

Hand

Approximately 90 % of hands with ULD have missing digits and 30 % have syndactyly [1]. Multiple digital anomalies can be seen with the hand ranging from a full complement of digits to just one digit. Ectrodactyly has been well documented in patients with ULD [1, 13, 15, 17]. Many of the existing digits are usually not normal, with variations of hypoplasia, missing phalanges or metacarpals, and variations of syndactyly and synostoses between phalanges and metacarpals [15].

Seventy percent of patients with ULD have abnormalities related to the thumb [1]. El Hassan et al. [17] reported that 11 of 17 limbs with ULD had digital anomalies, with four of those limbs having absent thumbs. Swanson et al. [11] and Broudy and Smith [21] reported that 68 and 100 % of their

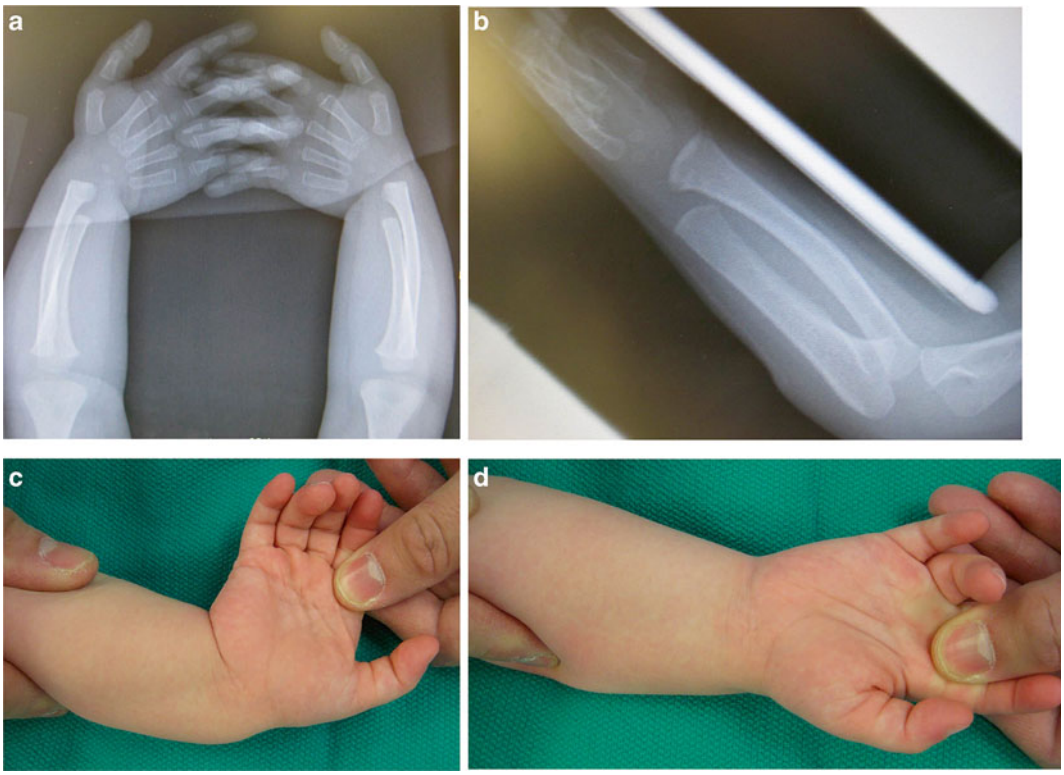

Fig. 10.1 (**a**) Anteroposterior and (**b**) lateral radiographs of a 2-year-old boy with bilateral ulnar longitudinal deficiency type II/A. (**c**) Clinical photograph showing excessive wrist ulnar deviation. (**d**) The wrist position rests in neutral

patients with ULD had radial-sided hand abnormalities, respectively. Cole and Manske [13] reported that 73 % of the 55 hands evaluated had an abnormal thumb or first web space. Their classification system describes the spectrum of thumb and first web space involvement from normal all the way to aplastic [13]. Evaluating the thumb and first web space deficiencies is important because surgical intervention to alter the radial-sided abnormalities in the hand may provide more substantial functional gains than operations elsewhere along the arm [1, 13, 15–17].

Treatment

Treatment of patients with ULD depends on the function of the limb. Non-operative intervention typically consists of early stretching and splinting starting at a young age. Depending on the function of the hand, surgical intervention may be warranted. Thus, the majority of surgical interventions in patients with ULD are performed on the hand, including syndactyly releases, deepening of the first web space, and first metacarpal rotational osteotomy [1, 13, 15, 16]. In special circumstances, other procedures including excision of the ulnar anlage, humeral rotational osteotomy, and creation of a one-bone forearm may be indicated.

Hand

Hand function can be improved with syndactyly releases, reconstruction of the thumb (opponensplasty, pollicization), and deepening of the first web space [1, 16, 23]. Ezaki and Carter [16] recommend delaying hand surgery until the second year of life. The reconstruction procedures of the hand are very important in improving the function of these children, and they recommend waiting for the hands to get larger to allow for a more precise surgery and thus better result [16].

First metacarpal rotational osteotomy is indicated in hands where the digits all lie in the same plane. The goal of rotation is to allow for prehension with the pulp of the digits. Rotation of other metacarpals and even phalanges to achieve this goal should be performed. Ezaki and Carter [16] report that there is a tendency for a slow loss of rotation after surgical intervention, and they recommend concomitant realignment of muscle power with tendon transfers to help prevent derotation.

Wrist

Controversy over excision of the ulnar anlage has been debated within the literature [11, 15–17, 21, 22]. However, there is some agreement as to which patient may benefit from early anlage excision. Indications for ulnar anlage excision [1, 15–17, 22] include the following:

1. Greater than 30° of fixed ulnar deviation
2. Clinically documented progression of ulnar deviation

It is recommended that excision of the ulnar anlage be performed at age 6 months. Proponents of early excision state it may improve both the function and appearance of the arm [1, 7, 22]. The anlage acts as a tether and will restrict radial growth and increase deformity of the forearm. In addition, the forearm will double in length by age 3 years, and resection of the anlage will provide the best possibility for unrestricted growth of the limb [16, 22].

To excise the ulnar anlage, either a longitudinal or lazy "S" incision is used over the ulnar border of the forearm and wrist. Usually the flexor carpi ulnaris is absent and the neurovascular bundle (if present) is directly under the skin and needs to be protected. Distally, it is crucial to dissect the anlage off of the carpus and radius completely. Following distal resection, the wrist should be passively corrected to a neutral position. Resection of the entire fibrous anlage proximally is not required, and usually resection of the distal third is adequate [16]. If excessive bowing of the radius is present, then an osteotomy can be performed at the same time. Postoperative course includes immobilization of the wrist in a neutral position for 6 weeks followed by stretching and splinting for at least 6 months, although some authors have recommended nighttime splinting with a short arm orthosis until skeletal maturity [23].

Forearm

The forearm in patients with ULD can be challenging to treat. Multiple procedures have been described: creation of a one-bone forearm [1, 15, 23, 24], radial osteotomies [18–20], and forearm lengthening [25, 26].

Several authors [1, 16, 27, 28] have advocated that the only indication for creating a one-bone forearm in patients with ULD is in the presence of forearm instability that is disabling. Thus, this procedure should rarely be done, knowing that any possible improvement in cosmetic appearance will be offset by the loss of function.

Radial osteotomies have been described [18–20] and may be performed at the same time of excision of the ulnar anlage if excessive bowing exists [16]. Although the forearm may be malrotated, most children do not require a forearm rotational osteotomy to improve their function [1].

Chen et al. [26] describe a case using an external fixator to gradually lengthen the ulna in a Bayne Type II. This patient also had a radial head dislocation. They reported an 81-mm lengthening over 7 months, with gradual reduction of the radial head. Elbow range of motion increased by 40° from 10° to 110° and preservation of preoperative forearm rotation was documented. This seems to be an option in this very specific subset of patients.

Elbow/Humerus

Rotational osteotomy about the elbow may be useful, especially in cases where the hand is positioned behind the child, as in the hand-on-flank deformity [1, 16–18, 23]. These patients typically have a radiohumeral synostosis with a hyperpronated forearm, bowing of the radius, and flexion and rotation of the elbow [17, 18]. The procedure can be performed at the level of the distal humerus through a lateral incision. Careful dissection is used to expose the humerus. Kirshner wires are placed distal and proximal to the proposed osteotomy site in a parallel fashion. The distal fragment is rotated so that hand is now positioned in front of the trunk. Care must be taken when performing the rotation, as serious damage to the vessels can occur. If needed, it is better to shorten the humerus as well. The osteotomy should be fixed with either transverse wires or plate and screws depending on the size of the patient. The arm can be protected in a long arm cast for 4–6 weeks.

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