Ulnar Longitudinal Deficiency

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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. 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 to 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 of children with ULD is variable. A majority will have involvement in their entire upper limb. Classification systems focus on the elbow/forearm abnormalities [7–11], hand [12], and more specifically the thumb and first web space [13].

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D. R. Laub Jr. (ed.), *Congenital Anomalies of the Upper Extremity*,
https://doi.org/10.1007/978-3-030-64159-7_11

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:

I. Normal length ulna with ulnar-sided hand anomalies
II. Hypoplasia of the ulna (presence of distal and proximal ulnar epiphysis)
III. Partial aplasia of the ulna (absence of the distal or middle one-third of the ulna)
IV. Total aplasia of the ulna (complete absence of the ulna)
V. 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 incorporating cases of severe radiohumeral synostosis with humeral bifurcation or a large medial epicondyle. Given the rarity of the disease along with variable presentation, Buck-Gramcko [15] stated that the pathological findings in ulnar deficiency are so different in their involvement and distribution that it was impossible for him to divide them into any classification system. Although Bayne and others describe the elbow and forearm abnormalities, treatment has really been focused more on the hand and digits. Cole and Manske [13] presented a classification system based upon the characteristics of the thumb and first web:

A. Normal first web space and thumb
B. Mild first web and thumb deficiency
C. 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
D. Absent thumb

The authors of this classification scheme point out that it is the complexity of the radial-sided problems that requires the majority of surgical procedures, so their classification will focus the surgeon's attention on those deficiencies most important for the restoration of function. Their conclusion was that ULD is best classified by an elbow/forearm system, supplemented by hand classification [13].

Associated Anomalies

Unlike patients with radial longitudinal deficiency, children with ULD rarely have heart or hematopoietic anomalies. However, these children 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

Patients with ULD may have hypoplasia of their proximal humerus and shoulder region. Despite this abnormality, most patients do not have restricted motion [15].

Elbow

There is quite a bit of variation in the clinical presentation of children with ULD. They may have an elbow that is stable, unstable, or fused. Their affected joints may have normal, hypoplastic, or severely deformed articular surfaces. Patients may present with congenital dislocation of the radial head, which may cause subsequent deformity to the distal end of the humerus [15]. El Hassan et al. [17] reported that 12% of the children they treated with ULD had a radiohumeral synostosis. In their series, they described patients' elbows in 20–90° of flexion and no elbows in full extension [17]. Others have described patients having elbows fixed in full extension [11] or with severe flexion and rotation, so that the hand is positioned behind the child and away from the opposite, uninvolved hand. This creates the so-called hand on flank deformity [18].

Forearm

Buck-Gramcko [15] reported that patients with different types of ulna defect showed no correlation to the severity of the involvement of other parts of the arm. He described patients presenting with ULD expressing all variations of other hand and elbow anomalies. Most patients with ULD will have a shorter than normal forearm (Fig. 11.1). Havenhill et al. [8] described a variation of patients with a normal forearm but deficiencies isolated to the ulnar side of the hand—a type 0 ulnar longitudinal deficiency.

In ULD, ulnar hypoplasia is most common (60%) with partial absence of the ulna reported in 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

Children may present with angulation of their wrist, but it is typically not as severe as that seen in radial longitudinal deficiency (see Fig. 11.1c). El Hassan et al. [17] reported that patients with ULD had wrists that were positioned in neutral in 71% of patients, with the remaining having 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 their wrists were in ulnar deviation, patients had limitations of radial deviation, wrist flexion, and extension [17]. Controversy over the role of the ulnar anlage and its relationship to wrist deviation

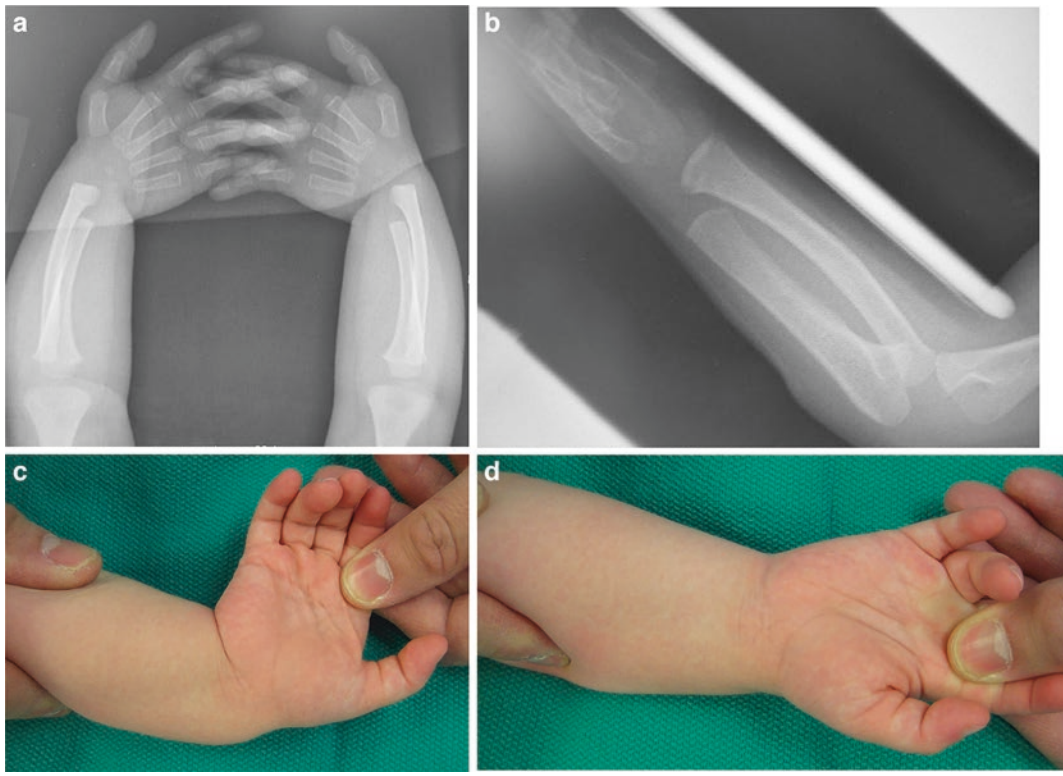

Fig. 11.1 (**a**) Anteroposterior and (**b**) lateral radiographs of a 2-year-old boy with bilateral ulnar longitudinal deficiency type II/A. (**c**) Clinical photograph showing excessive wrist ulnar deviation. (**d**) The wrist position rests in neutral

continues [15–17]. Patients may present with absent carpal bones in correlation with missing digital rays, and synostoses occur in 30% to 40% of cases [15].

Hand

Approximately 90% of patients with ULD have missing digits and 30% have syndactyly [1]. Multiple digital anomalies can be seen in the patients with ULD, ranging from a full complement of digits to just one digit. Ectrodactyly has been well documented in patients with ULD [1, 13, 15, 17]. Often, the patient's existing digits are not normal, with variations of hypoplasia, missing phalanges or metacarpals, 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 patients with ULD had radial-sided hand abnormalities, respectively. Cole and Manske [13] reported that 73% of the 55 patients 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 a patient's thumb and first web space deficiencies is important, as surgical intervention to alter the radial-sided abnormalities in the hand may provide more substantial gains for a patient's function than operations elsewhere along their arm [1, 13, 15–17].

Treatment

Treatment of patients with ULD depends on the function of the limb. Nonoperative 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. Tissue distraction is a more invasive method of stretching and may be an adjunct to surgical management. 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 osteotomies [1, 13, 15, 16]. In special circumstances, other procedures, including excision of an 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 child's second year of life. The reconstruction procedures of a child's hand are very important in improving their function; waiting for the child's hands to get larger allows for a more precise surgery and thus a better result [16].

First metacarpal rotational osteotomy is indicated when a child's hands has digits that all lie in the same plane. The goal of this rotation is to allow for prehension with the pulp of the digits. Rotation of other metacarpals and even phalanges to achieve this goal should also be considered. 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 continues to be debated within the literature [11, 15–17, 21, 22]. However, there is some agreement as to which patients 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 a patient's 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 will need to be protected. Distally, it is crucial to dissect the anlage off of the carpus and radius completely. Following distal resection, the patient's wrist should be passively corrected to a neutral position. Resection of the entire fibrous anlage proximally is not required; 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 management includes immobilization of the patient's wrist in a neutral position for 6 weeks followed by stretching and splinting for at least 6 months. Some authors have recommended nighttime splinting with a short arm orthosis until patients reach skeletal maturity [23].

Forearm

The forearm of 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 is in the presence of forearm instability that is disabling to the patient. 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 as 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 distraction osteogenesis of the ulna in a child with a Bayne type II deformity. They reported an 81-mm lengthening over 7 months, with gradual reduction of a dislocated radial head. Elbow range of motion increased and preservation of preoperative forearm rotation was documented.

Schachinger et al. described the use of soft tissue distraction in two children with Bayne type II deformities prior to definitive one-bone forearm surgery [29]. This seems to be an option in this very specific subset of patients.

Elbow/Humerus

When a child's hand is positioned behind the body, the "hand-on-flank deformity," a rotational osteotomy near their elbow may be useful [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 skeletal fragment is rotated so that the patient's hand is now positioned in front of the trunk. Care must be taken in this rotation surgery, as the patient's vessels and nerve are at risk for serious damage. If needed, it may be useful to shorten the patient's humerus as well. The osteotomy can be fixed with either transverse wires or plate and screws depending on the size of the patient. Their arm can then be treated postoperatively in a long arm cast for 4–6 weeks.

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