Radial Longitudinal Deficiency: Radius Hypoplasia

7

Chris Stutz and Scott Oishi

Introduction

Radial longitudinal deficiency (RLD) comprises a spectrum of clinical manifestations involving phenotypic changes of the upper extremity that range from underdevelopment to complete absence of the radial sided structures. The majority of cases of RLD are sporadic in occurrence, but the deformity can be passed genetically as well. Treatment for this condition varies depending on the clinical presentation of the patient as well as any associated anomalies that may exist. This chapter will attempt to explain the background and etiology of RLD, outline the conditions that have been associated with the deformity, review the classification of the various phenotypic presentations, and review current treatment patterns and their associated outcomes.

Background

The first documented case of RLD, then termed "radial club hand" was reported by Petit in 1733 when he described the findings in an infant autopsy. The term "radial club hand" has been largely supplanted in the modern literature with the term "radial longitudinal deficiency." In 1894, Sayre published the first case of RLD treated with centralization to address the radial deviation deformity associated with the condition by outlining the steps of centralizing the carpus on the end of the distal ulna. Since the time of these early publications, there have been significant advances in the understanding of the diagnosis, the deformity, and its associated conditions. Despite these advances, there remains little consensus in opinion regarding the best operative or non-operative treatment of the radial deformity in children with RLD.

Etiology

The theories regarding the embryologic basis for RLD continue to evolve, as the specific mechanisms of limb bud development are uncovered. In animal models, the progressive reduction of apical ectodermal ridge associated fibroblastic growth factors causes a progressive reduction in the size and volume of the developing limb bud. These alterations in cellular communication result in deformities that resemble those seen clinically in RLD [1, 2]. Mutagenic agents given to pregnant rats at various time points in gestation resulted in a substantial portion of littermates exhibiting manifestations consistent with RLD. The manifestations correlated with the time of administration and the dose of the mutagenic agent [3]. The prevalence of RLD has been reported as 1 in 55,000 live births [4], with a male to female ratio of 3:2 [5–8].

Associated Conditions

The association of RLD with certain medical conditions is well established. Historically, patients diagnosed with RLD were given a poor general prognosis, likely related to the associated morbidity of the related medical conditions [9]. Goldfarb et al. [10] reported on 164 patients with RLD, 67 % of which had associated medical or musculoskeletal abnormalities. The investigators reported the relative incidence of associated medical conditions was directly related to the severity of the RLD, with the most common related conditions being cardiac anomalies (20 %), thrombocytopenia-absent radius syndrome (15 %), VACTERL association (13 %), Holt-Oram syndrome (4 %), and Fanconi anemia (1 %).

C. Stutz, M.D.
Department of Orthopaedics, Monroe Carell Jr Children's Hospital, Vanderbilt University Medical Center, 4202 Doctors' Office Tower, 2200 Children's Way, Nashville, TN 37204, USA
e-mail: Chris.Stutz@Vanderbilt.edu

S. Oishi, M.D. (✉)
Hand Service, Texas Scottish Rite Hospital for Children, 2222 Welborn Street, Dallas, TX 75219, USA
e-mail: scottoishi@aol.com; scott.oishi@TSRH.org

D.R. Laub Jr. (ed.), *Congenital Anomalies of the Upper Extremity: Etiology and Management*,
DOI 10.1007/978-1-4899-7504-1_7, © Springer Science+Business Media New York 2015

Hall et al. defined thrombocytopenia-absent radius as a syndrome in 1969 [11]. The inheritance pattern was thought to be autosomal recessive, but reports of parent-to-child transmission and multiple affected relatives in families suggest either heterogeneity or a different mode of inheritance [12–14]. Further genetic investigations have found a specific microdeletion of chromosome 1q21.1, which is necessary, but in itself insufficient to cause the thrombocytopenia-absent radius phenotype [15]. The cardinal findings of TAR syndrome are the absence of radii with the presence of hypoplastic thumbs and thrombocytopenia [16]. The presence of an aberrant muscle, termed the brachiocarpalis, was identified by Oishi and colleagues in the upper extremities of children with TAR syndrome contributing to the radial angulation deformity of the carpus [17]. Unique to this diagnosis is that even though the thrombocytopenia can initially be severe, it usually spontaneously resolves over time without the need for intervention.

VACTERL association is a nonrandom association of birth defects involving vertebral anomalies, anal atresia, cardiovascular anomalies, trachea-esophageal fistula, renal and/or radial anomalies, and limb defects. VACTERL association is likely related to multiple factors, but can be seen with chromosomal defects such as Trisomy 18 and is encountered more commonly in children of diabetic mothers [18]. There has been no specific genetic cause identified in VACTERL association to date. RLD patients must have at least three, including RLD, of the possible associations to be considered a VACTERL patient.

Holt-Oram syndrome is an autosomal dominant condition hallmarked by cardiac abnormalities and upper limb anomalies involving the radial ray. The genetic abnormality responsible for the syndrome has been identified as a missense mutation in the TBX5 gene [19]. The upper extremity involvement in Holt-Oram is variable. There is commonly hypoplasia of the radial elements with or without bizarre synostoses between the radius and ulna (Fig. 7.1).

Fanconi anemia is the most common inherited cause of bone marrow failure [20]. The bone marrow failure most commonly occurs between the ages of 5 and 15. Phenotypic variations are common in presentation and include short stature, thumb and radius deformities, hyperpigmentation of skin, renal, cardiac, and genitourinary abnormalities [21]. The diagnosis can be made using a chromosome breakage analysis (diepoxybutane analysis). The test is expensive and its use as a routine screening tool in patients with apparent isolated RLD continues to be debated. However, the advent of successful pediatric bone marrow transplantation has led some authors to feel that diepoxybutane testing is important in every child with an RLD diagnosis.

Unique to many other conditions treated by the discipline of hand surgery, RLD often offers the hand surgeon the opportunity to be the first to make a diagnosis of other associated anomalies. This is related to the fact that the visible difference in upper extremity development often implores the parents and pediatrician to pursue evaluation for treatment of the affected limb. Hence, it is imperative that the hand surgeon be aware of these common associations and performs a complete evaluation of the child in all cases. This evaluation should include, at a minimum, a complete musculoskeletal and systemic evaluation, a complete blood count, echocardiogram, abdominal ultrasound, and subsequent evaluation for scoliosis.

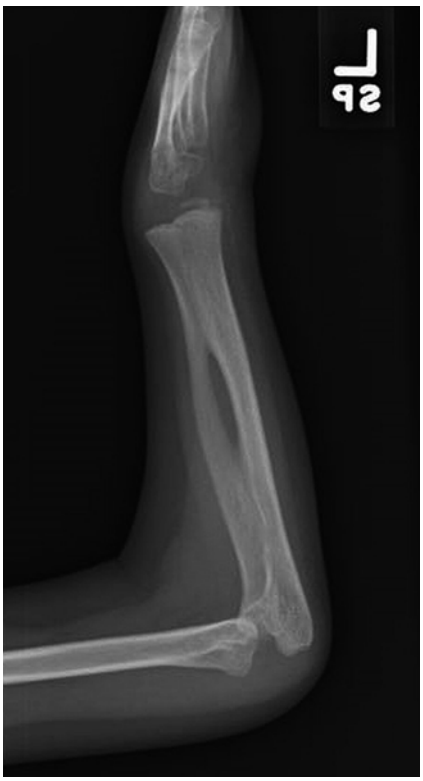

Fig. 7.1 Bizarre forearm synostosis in a patient with Holt-Oram syndrome

Classification

The original classification of RLD was described by Bayne and Klug in 1987 [22]. They based the classification system on the radiographic appearance of the radius and divided the phenotype into four categories. Type I was defined as a short radius with delayed appearance of the distal radial epiphysis. Type II was defined as a "radius in miniature" with growth of both proximal and distal radial epiphyses affected. Type III denoted partial absence of the radius with no distal radial physis; Type IV was defined as complete absence of the radius.

The original classification of scheme of Bayne and Klug was modified by James et al. [23] in 1999 to include Types N and 0 with further delineation of what constituted Type I

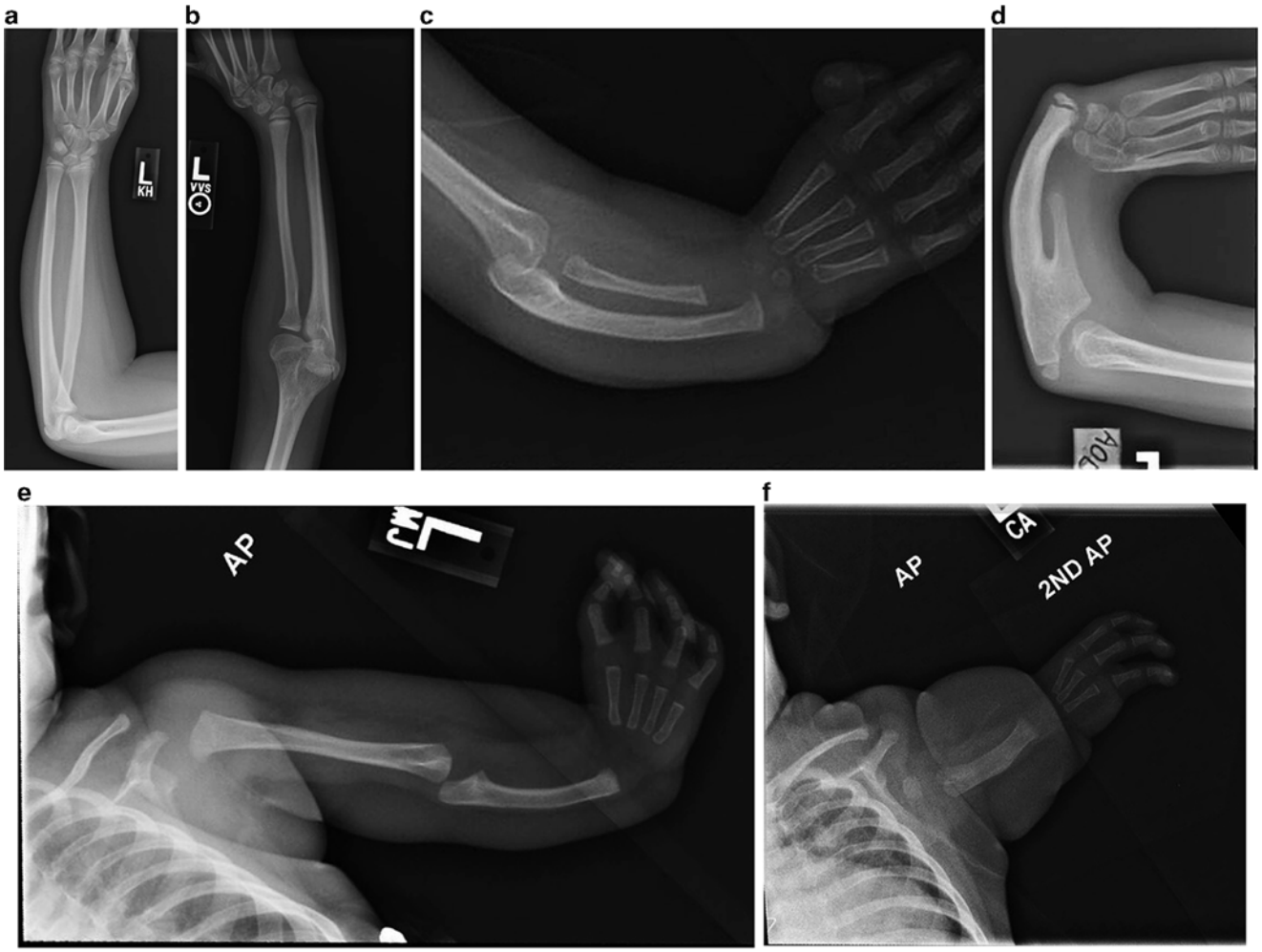

Fig. 7.2 (**a**) Type 0/N. (**b**) Type I. (**c**) Type II. (**d**) Type III. (**e**) Type IV. (**f**) Type V

RLD. The classification was further modified by Goldfarb et al. [24] in 2005 to include more severe proximal manifestations of RLD as Type V. The current state of RLD classification is as follows (Fig. 7.2):

Type N—The thumb is hypoplastic or absent in the presence of a normal carpus or radius. Radial angulation at the wrist is usually absent or minimal.

Type 0—The radius is of normal length with proximal and distal physes. The radial carpal bones are hypoplastic or absent. The degree of radial angulation of the wrist is variable. The angulatory deformity is owing to the abnormal carpal bones and the presence of tight soft tissue structures on the radial side of the wrist, including the wrist capsule and musculotendinous structures.

Type I—The radius is foreshortened by at least 2 mm compared to the distal ulna. The distal radial physis is present but its growth is slowed. The proximal radial physis is present and of normal morphology. Radio-ulnar synostosis or congenital radial head dislocation is variably present.

Type II—The radius is hypoplastic in its entirety with proximal and distal physes present—the so-called radius in miniature. This can be associated with notable ulnar bowing.

Type III—The distal portion of the radius is absent. There is no distal radial physis.

Type IV—The radius is absent in its entirety. This is the most common phenotypic presentation of RLD [22].

Type V—This represents a severe proximal form of RLD formerly considered phocomelia. Taking into account principles of developmental biology, the concept of a true intercalary defect has been challenged by recent authors [24, 25]. Extremities in this category have an abnormal glenoid, absence of the proximal portion of the humerus, articulation of the distal humerus with the ulna, and radial sided hand abnormalities.

Clinical Presentation

While the etiology of the condition hinges on the longitudinal dysplasia of the radius, the clinical presentation of patients with radial longitudinal dysplasia is diverse. Patients can often present with skeletal abnormalities that extend beyond the radial deficiency. These include shortening of the forearm and/or bowing of the ulna, absent or limited elbow flexion, and absence or hypoplasia of the scaphoid and other carpal bones. Thumb hypoplasia can be present and consist of hypoplasia of the thenar intrinsic and/or extrinsic musculature, hypoplasia of the skeletal elements with or without associated articular instability, rudimentary presence of the thumb ("pouce floutant"), or complete absence of the thumb. The fingers can exhibit limited flexion, with the radial digits more affected than the ulnar digits. In addition to the manifestations of RLD in the hand, the soft tissues on the radial side of the wrist and forearm are tight contributing to the radial angulation of the hand plate on the distal ulna. The extrinsic wrist extensors are often poorly developed, and the malformed radial soft tissues often form a fibrous tether to the radial side of the wrist. This combination results in the classic presentation of a radial deviated wrist held in a flexed posture.

The abnormalities have both aesthetic and functional consequences. In severe cases the appearance of the extremity can be unsightly secondary to the shortened forearm and the angled, flexed posture of the wrist and hand. On average, the forearm length is 54 % of normal, ranging from 37 to 67 % [26]. This limits the extremity's reach and can make two-handed activities with the normal, opposite extremity difficult. In patients with bilateral upper extremity involvement, the functional limitations can be more severe. James et al. [23] found the incidence of bilateral involvement to be 65 % in a study of 104 patients. If poorly functioning digits are present, this can further impede function. Unfortunately, when present, finger dysfunction is rarely amenable to surgical correction. This is in contradistinction to thumb limitations, where several options are available to improve function.

Non-operative Management

The non-operative care of a child with RLD often begins very early in life. Occupational therapy intervention is commonly instituted during the first few weeks of life, especially if the infant requires hospitalization for associated abnormalities. Those children whose health allows them to be discharged from hospital care in the first few days of life are often referred for outpatient therapy services very early on by their pediatricians.

Therapeutic intervention at this point includes stretching exercises aimed at lengthening the contracted tissues on the radial side of the wrist and improving the hand-forearm angle. Splinting is often used as an adjunct to stretching in an effort to maintain the wrist in the corrected position and provide static resistance to a resting position of radial deviation. Specific therapeutic protocols for treatment of RLD by non-operative means vary widely from surgeon to surgeon and therapist to therapist. There have been no published reports of therapeutic regimens proven to change the natural history of RLD, although its effectiveness in teaching children to use the affected limb in an efficient and useful manner has been seen clinically by many who care for these patients. Timing of intervention is also a topic of debate among those who treat these children. The authors feel that an early stretching regimen with nap and night splinting can be instituted early in life, but the parents should be encouraged to remove the splints for extended periods while the child is awake to allow him/her to interact appropriately with his/her surroundings and obtain the sensory interaction with the environment that is essential for proper development. Two-handed activities generally begin around the age of 3 months. At this time, splint wear during awake hours may become beneficial to place the hand in a less radially deviated position, functionally increasing the length of the affected extremity, and allowing for easier two-handed manipulation of objects.

Operative Management

There have been many procedures described for the management of the wrist and forearm deformity in RLD. Since the original description of centralization by Sayre in 1894, several authors have published similar techniques with slight variations to the original procedure [27–30]. In addition, newer techniques such as radialization, pre-centralization distraction, and microsurgical transfer of vascularized epiphyses have been introduced to treat the deformity [31–34]. No single procedure has proven superior to another. Hence there remains vast disparity in treatment recommendations between surgeons treating the condition. Recurrence of the radial angulation remains the Achilles heel for procedures aimed at correcting the deformity [35].

Reports centered on treatment of Types 0, N, I, and II RLD are sparse.

Type 0

Despite the relative frequency of Type 0 RLD reported by James et al. [23], a small number of these patients require surgical intervention. In 2004, Mo and Manske [36] reported on six wrists in five children treated with surgical correction.

They recommended surgical intervention for radial deviation deformity greater than 20°. In their subset of patients, the preoperative hand-forearm angle ranged from 35° to 70° with all wrists lacking active extension to neutral. The authors describe a dorsal approach to the wrist with exposure of the extensor carpi radialis tendon or tendons. The tendon is released from its distal insertion. Following release, the dorsal–radial wrist capsule, as well as the volar wrist capsule, is released allowing passive correction of the wrist to neutral position. The extensor carpi ulnaris tendon is released, leaving a distal stump for tenoraphy with the radial wrist extensors, effectively removing the radial deviation force and realigning it to gain neutral wrist extension. The proximal stump of the extensor carpi ulnaris tendon is sewn into the dorsal wrist capsule overlying the third metacarpal to further augment active wrist extension. Optionally, a pin can be placed across the carpus into the distal ulna to maintain the wrist in its corrected position. The patient is then casted in neutral to slight wrist extension for 6–8 weeks. The cast and pin, if present, are removed and the patient is allowed to begin active range-of-motion exercises. At rest the patient is splinted in the corrected position for an extended duration.

Mo and Manske [36] reported favorable outcomes using the above surgical technique. They reported an average improvement of radial deviation at rest from 58° to 12°, with active wrist extension improving an average of 53° and passive wrist extension improving and average of 28°. The average length of follow-up was 19 months (range, 2–38 months).

Types I and II

There have been few published reports on the treatment of Types I and II radial longitudinal deficiencies. Often, children with these types of RLD do not require surgical intervention. When necessary, the most common form of treatment is radial lengthening with release of the tight radial soft tissues and tendon transfer to support the realigned position. Lengthening of the radius is most commonly done by way of osteotomy and lengthening through an external fixator [37–40]. Others have reported on lengthening of the radius acutely, with gains of up to 1.6 cm [41]. Many authors have described techniques of lengthening through an external fixator with slight variations. Depending on surgeon preference, the lengthening can be performed with a single plane fixator [38] or by using a ring-type fixator [40]. When performing acute radius lengthening, Waters et al. [41] described a technique of using a temporary external fixator intraoperatively for distraction of the radius after performing a Z-cut osteotomy, followed by plate fixation of the bone in its new lengthened position.

Matsuno et al. [38] reported on two patients with Type II RLD who underwent radial lengthening with an external fixator. The outcomes demonstrated recurrence of the deformity following fixator removal with and increase the hand-forearm angle at final follow-up.

Types III and IV

The treatment of Types III and IV RLD is classically described as centralization of the carpus on the distal end of the ulna. Since Sayre first described the original procedure of centralization in 1894, multiple authors have published their experience using this technique, as well as several modifications to the procedure aimed at decreasing the recurrence of the radial angulation deformity. In addition, many others have suggested alternative procedures to accomplish the task of neutralizing the carpus on the end of the forearm. These procedures include radialization of the carpus, transfer of vascularized epiphyses to support the radial side of the carpus, and ulnocarpal fusion [31, 34, 42, 43].

Centralization

The centralization procedure is based on four surgical steps: (1) initial stretching of soft tissues ± pre-centralization distraction, (2) surgical alignment of the carpus on the ulna, (3) balancing of the deforming forces, and (4) maintenance of the corrected position.

Historically, stretching of the radial tissues was accomplished by serial cast application prior to surgical centralization, often carried out within the first several months of life. This technique fails to adequately distract the tight radial soft tissues or translate the carpus distally over the end of the ulna; instead it simply aligns the carpus alongside the distal ulna. In addition, the early application of casts precludes the use of the extremity by the child during the formative time of "learning" single and two-handed object manipulation. As a result, the use of external fixation to accomplish soft tissue distraction has been advocated in recent years by some surgeons. The application of uniplanar [44], biplanar [32, 45], and ring [33, 46, 47] external fixators have been described. The use of external fixation allows for the correction of the radial deviation deformity through distraction of the radial soft tissues and correction of the volar subluxation of the carpus in relation to the distal ulna. Distraction of the deformity is begun 3–5 days following the application of the fixator. The distraction is carried out at a rate of 0.5–1 mm per day until the desired position of the carpus is accomplished. The extremity is then maintained in the fixator for a period of 3 to 4 weeks prior to surgical stabilization of the carpus in its centralized position to allow the soft tissues to equilibrate.

Originally, the centralization procedure was performed through a longitudinal dorsal incision. Since that time, there have been multiple incisional techniques described to accomplish surgical centralization of the carpus [27, 28, 48].

The pre-centralization distraction of the soft tissues allows for ease in accomplishing surgical centralization while often obviating the need for transposition flaps for soft tissue coverage. Regardless of the incision used, the hypoplastic extensor tendons are carefully identified and retracted. The tight dorsal, radial, and volar wrist capsule and soft tissues are released to allow for a tension-free placement of the carpus onto the distal ulna aligned on the axis of the third metacarpal. Buck-Gramcko described "radialization" of the carpus in which he aligned the carpus on the axis of the second metacarpal in an effort to decrease the tendency towards recurrence of the deformity [31]. With the use of preoperative distraction, the need for "notching" [49] of the carpus to decrease soft tissue tension is usually unnecessary. The importance of obtaining a tension-free centralization has been reinforced by Sestero and Van Heest [50], who demonstrated that ulna in non-centralized radial longitudinal deficient extremities attained 64 % of normal length while the ulnar length in centralized extremities was 58 % of normal compared to 48 % of normal when notching of the carpus was performed. They postulated that the decrease in longitudinal growth capacity of the ulna was secondary to increased pressure applied to the distal ulnar physis by the centralized carpus. Once an appropriate centralized position is obtained, carpus is pinned to the ulna with longitudinal Kirschner wires (K-wires) taking care to avoid the distal ulnar physis. The pins are cut beneath the skin and often remain in place for up to 6 months postoperatively to maintain the corrected position. Soft tissue rebalancing procedures are then performed to redirect the forces across the centralized carpus. The extensor carpi ulnaris tendon is advanced to improve the ulnar and dorsal vector of pull to the wrist and hand [22, 30, 31]. If present, the radial wrist extensors are transferred ulnarly to alleviate the deforming force caused by their function. The digital extensors are translated in an ulnar direction using a sling of extensor retinaculum to align them along the longitudinal axis of the ulna, hence eliminating another deforming force.

Epiphyseal Transfer

The concept of supporting the hand and carpus by transferring bony elements to the radial side of the wrist to augment the support provided by the distal ulna was introduced in 1928 by Albee [51] and attempted by several subsequent authors [6, 8, 29]. Unfortunately, these early attempts were hindered by the limited growth potential possessed by the transferred nonvascularized tissue. With the advent and refinement of microsurgical techniques, the concept of vascularized epiphyseal transfer with retained growth potential [52–54] rejuvenated the interest in supporting the radial side of the carpus using a structural graft. In 1998, Vilkki [34] reported on the use of the second metatarsophalangeal joint to support the radial side of the carpus.

In contrast to the centralization procedure, the epiphyseal transfer is generally performed at an age of 4–5 years. Prior to embarking on the microsurgical portion of the reconstruction, the child often undergoes a soft tissue release with detethering of the radial side of the carpus with concomitant volar bilobed flap, transposing the excess ulnar sided soft tissue to the deficient radial side [55]. This early intervention (done at approximately 12–18 months of age) has the advantage of maintaining wrist motion while minimizing risk to the distal ulnar physis. Following release and soft tissue transfer, a protocol of stretching and splinting is maintained through the early childhood years in an effort to preserve the increase in motion.

At an age of 5–6 years, the child is evaluated for the possibility of microsurgical epiphyseal transfer. Often the child and his family decline additional surgery because very few functional limitations exist and cosmesis would be the primary indication for surgery. That said, if further surgical reconstruction is warranted, the microsurgical epiphyseal transfer is preceded by soft tissue distraction using an external fixator as described earlier in the chapter. The frame is applied and the carpus is slowly distracted (0.5–1 mm per day) until the desired anatomic position of the hand is accomplished over the distal ulnar. This can take 6–8 weeks to accomplish. The second toe metatarsophalangeal joint is harvested from the ipsilateral limb maintaining two arterial sources—first and second dorsal metatarsal artery and second and third plantar metatarsal artery [56]. Flexor and extensor tendons are preserved and sutured to the remaining proximal phalanx. The dorsal cutaneous nerves are also preserved to the dorsal skin paddle. The middle and distal phalanges of the toe are excised. Exquisite care must be taken to preserve the vessels to the epiphysis of the proximal phalanx and metatarsal during harvest.

The metatarsophalangeal joint is transferred to the wrist through a dorsal ± volar incision. The metatarsal is anchored to the ulna using K-wires, which are cut and bent beneath the skin. The proximal phalanx is anchored to the base of the second metacarpal, or against the scaphoid if present, in a position of 15–20° of flexion to increase stability. The preserved tendons of the toe are then sutured to the radial flexor and extensor tendons or muscle bellies to confer additional stability. After securing the bony construct, the metatarsophalangeal joint is revascularized.

Oftentimes the radial artery is absent in limbs affected by RLD; hence, the arterial supply for the epiphyseal transfer is provided by a persistent median artery or the ulnar artery. If present, the median artery or radial artery is anastomosed to the dominant vessel of the metatarsophalangeal joint in end-to-end fashion. In those cases where the median and radial artery is absent, the dominant vessel of the metatarsophalangeal joint is anastomosed to the ulnar artery in end-to-side

fashion. Following acquisition of arterial inflow, the venous drainage is accomplished by anastomosis of dorsal veins.

The distraction device and K-wires are removed after radiographs have confirmed bony consolidation, usually 6–8 weeks. The arm is then casted for an additional month to protect the maturing transfer.

Ulnocarpal Arthrodesis

Ulnocarpal arthrodesis [43], or epiphyseal ulnocarpal arthrodesis [42] for the skeletally immature, is the procedure that most effectively stabilizes the wrist and improves the appearance of the radial angulation deformity. Despite the improvement in appearance, some have questioned the benefit of arthrodesis citing the maintenance of wrist motion as a substantial benefit in the function of the radial deficient limb [7]. Hence, the procedure is often thought of as a salvage procedure for severe, recurrent deformity. Rayan reported on two cases of recurrent deformity in skeletally mature patients who underwent ulnocarpal arthrodesis with improvement in both appearance and function [43]. Pike et al. [42] reported on 12 post-centralization wrists treated with ulnocarpal epiphyseal arthrodesis for recurrent radial angulation >45° and/or inability to extend the wrist beyond 25°. Post-operatively, the wrists were stable at an average of 20° radial angulation and 11° of flexion. All reported improvement in appearance and function post-operatively. A trial of ulnocarpal pinning can be considered for patients/parents who have concern regarding postoperative function prior to performing definitive arthrodesis procedure.

Distraction-Lengthening of the Ulna

In order to address the functional limitation of impaired "reach" of the affected extremity, authors have reported lengthening of the ulna using a ring or uniplanar external fixator in several small series ranging from 4 to 9 patients [39, 57–59]. The distraction time ranged from 11 to 15 weeks, followed by a 23–32-week consolidation period. Average length gained in each extremity was 4.4–6 cm (46–54 % of total length). Complications of lengthening included callus fracture, delayed union, digital and wrist stiffness, pain, pin tract infection, and recurrence of radial angulation. There were no rigid outcomes reported documenting improvement in function of the lengthened extremity.

Outcomes/Complications

Regardless of the type of surgery utilized, the common denominator in the outcomes of the surgical management of RLD is the recurrence of the radial angulation deformity. Multiple studies have documented the recurrence of radial angulation deformity following centralization [26, 35, 60]. An average radial-forearm angle of 21–26° immediately after centralization has been noted in these studies, with an additional 9- to 38-degree increase in radial angulation occurring over time. The avoidance of recurrent deformity has not been alleviated by the use of pre-centralization distraction as shown by Dana et al. [61]. In 2008, Vilkki [62] presented the long-term study of 19 wrists treated with microsurgical epiphyseal transfer with an average of 11 years follow-up. The average hand-forearm angle was 28° of radial deviation with mean total active wrist motion of 83°. Of the nine wrists included in his original report [34], seven were noted to have increased radial angulation (mean of 12°) over a follow-up period of 15.2 years. Goldfarb et al. [26] reported significant functional limitations of the post-centralized hand, noting a 62 % increase in the Jebsen-Taylor timed activity tests compared to normal. Interestingly, the DASH scores showed only mild functional limitation. Buffart et al. [63] observed grip and pinch strength values of 36 % and 30 %, respectively, when compared to normal controls. Both of the previous studies represent post-centralization scores compared to normal controls. There are no comparisons to pre-centralization function, thus making it impossible to determine the effects of surgical deformity correction.

Complications of the surgical treatment of radial deficient limbs are both all inclusive and dependent of the surgical technique used. Recurrence of deformity is a complication that is ubiquitous despite the treatment modality. Pin tract infections, callus fracture, delayed union, and stiffness are common to all techniques utilizing external fixation. Damage to the distal ulnar physis, further impairing its ability to accomplish longitudinal growth, is the most feared complication of centralization. Hence, the concept of carpal notching has been largely supplanted by newer techniques of pre-distraction centralization, in an effort to diminish the forces exerted across the distal ulnar physis.

Future Directions

The best treatment of RLD and its multiple phenotypes remains a popular topic among surgeons commonly treating the condition. To date, treatment algorithms have encompassed the full circle of management strategies, from non-operative to operative care at various stages of life for various clinical presentations utilizing a vast array of surgical procedures. Certainly, the definitive "best" treatment has yet to be determined, and likely is not the same for every patient. Future comparisons of those treated for RLD with surgical intervention versus those treated by nonoperative means may shed the most meaningful light on what interventions benefit these children the most.

References

1. Mariani FV, Ahn CP, Martin GR. Genetic evidence that FGFs have an instructive role in limb proximal-distal patterning. Nature. 2008;453(7193):401–5. doi:10.1038/nature06876. Epub 2008/05/02. PubMed PMID: 18449196; PubMed Central PMCID: PMC2631409.
2. Sun X, Mariani FV, Martin GR. Functions of FGF signalling from the apical ectodermal ridge in limb development. Nature. 2002; 418(6897):501–8. doi:10.1038/nature00902. Epub 2002/08/02. PubMed PMID: 12152071.
3. Kato H, Ogino T, Minami A, Ohshio I. Experimental study of radial ray deficiency. J Hand Surg Br. 1990;15(4):470–6. Epub 1990/11/01. PubMed PMID: 2269841.
4. Lourie GM, Lins RE. Radial longitudinal deficiency. A review and update. Hand Clin. 1998;14(1):85–99. Epub 1998/04/04. PubMed PMID: 9526159.
5. Flatt A. Radial clubhand. The care of congential hand anomalies. 2nd ed. St. Louis: Quality Medical Publishing; 1993. p. 366–410.
6. Heikel HV. Aplasia and hypoplasa of the radius: studies on 64 cases and on epiphyseal transplantation in rabbits with the imitated defect. Acta Orthop Scand. 1959;39:1–155. PubMed PMID: 14400621.
7. Lamb DW. Radial club hand. A continuing study of sixty-eight patients with one hundred and seventeen club hands. J Bone Joint Surg. 1977;59(1):1–13. PubMed PMID: 833156.
8. Riordan DC. Congenital absence of the radius. J Bone Joint Surg. 1955;37-A(6):1129–39; discussion, 39–40. PubMed PMID: 13271460.
9. Kelikian H. Radial ray defect. Congenital deformities of the hand and forearm. Philadelphia: WB Saunders Company; 1974. p. 780–824.
10. Goldfarb CA, Wall L, Manske PR. Radial longitudinal deficiency: the incidence of associated medical and musculoskeletal conditions. J Hand Surg. 2006;31(7):1176–82. PubMed PMID: 16945723.
11. Hall JG, Levin J, Kuhn JP, Ottenheimer EJ, van Berkum KA, McKusick VA. Thrombocytopenia with absent radius (TAR). Medicine. 1969;48(6):411–39. Epub 1969/11/01. PubMed PMID: 4951233.
12. Ward RE, Bixler D, Provisor AJ, Bader P. Parent to child transmission of the thrombocytopenia absent radius (TAR) syndrome. Am J Med Genet Suppl. 1986;2:207–14. Epub 1986/01/01. PubMed PMID: 3146292.
13. Schnur RE, Eunpu DL, Zackai EH. Thrombocytopenia with absent radius in a boy and his uncle. Am J Med Genet. 1987;28(1):117–23. doi:10.1002/ajmg.1320280117. Epub 1987/09/01. PubMed PMID: 3314504.
14. Edelberg SB, Cohn J, Brandt NJ. Congenital hypomegakaryocytic thrombocytopenia associated with bilateral absence of the radius—the TAR syndrome. Hum Hered. 1977;27(2):147–52. Epub 1977/01/01. PubMed PMID: 863461.
15. Klopocki E, Schulze H, Strauss G, Ott CE, Hall J, Trotier F, et al. Complex inheritance pattern resembling autosomal recessive inheritance involving a microdeletion in thrombocytopenia-absent radius syndrome. Am J Hum Genet. 2007;80(2):232–40. doi:10.1086/510919. Epub 2007/01/20. PubMed PMID: 17236129; PubMed Central PMCID: PMC1785342.
16. Toriello HV. Thrombocytopenia-absent radius syndrome. Semin Thromb Hemost. 2011;37(6):707–12. doi:10.1055/s-0031-1291381. Epub 2011/11/22. PubMed PMID: 22102274.
17. Oishi SN, Carter P, Bidwell T, Mills J, Ezaki M. Thrombocytopenia absent radius syndrome: presence of brachiocarpalis muscle and its importance. J Hand Surg. 2009;34(9):1696–9. PubMed PMID: 19773129.
18. Stevenson RE, Hunter AG. Considering the embryopathogenesis of VACTERL Association. Mol Syndromol. 2013;4(1–2):7–15. doi:10.1159/000346192. Epub 2013/05/09. PubMed PMID: 23653571; PubMed Central PMCID: PMC3638783.
19. Huang T. Current advances in Holt-Oram syndrome. Curr Opin Pediatr. 2002;14(6):691–5. Epub 2002/11/19. PubMed PMID: 12436037.
20. Shimamura A, Alter BP. Pathophysiology and management of inherited bone marrow failure syndromes. Blood Rev. 2010;24(3):101–22. doi:10.1016/j.blre.2010.03.002. Epub 2010/04/27. PubMed PMID: 20417588; PubMed Central PMCID: PMC3733544.
21. Soulier J. Fanconi anemia. Hematology Am Soc Hematol Educ Program. 2011;2011:492–7. doi:10.1182/asheducation-2011.1.492. Epub 2011/12/14. PubMed PMID: 22160080.
22. Bayne LG, Klug MS. Long-term review of the surgical treatment of radial deficiencies. J Hand Surg. 1987;12(2):169–79. PubMed PMID: 3559066.
23. James MA, McCarroll Jr HR, Manske PR. The spectrum of radial longitudinal deficiency: a modified classification. J Hand Surg. 1999;24(6):1145–55. PubMed PMID: 10584934.
24. Goldfarb CA, Manske PR, Busa R, Mills J, Carter P, Ezaki M. Upper-extremity phocomelia reexamined: a longitudinal dysplasia. J Bone Joint Surg. 2005;87(12):2639–48. PubMed PMID: 16322613.
25. Tytherleigh-Strong G, Hooper G. The classification of phocomelia. J Hand Surg Br. 2003;28(3):215–7. PubMed PMID: 12809650.
26. Goldfarb CA, Klepps SJ, Dailey LA, Manske PR. Functional outcome after centralization for radius dysplasia. J Hand Surg. 2002;27(1):118–24. PubMed PMID: 11810625.
27. Evans DM, Gateley DR, Lewis JS. The use of a bilobed flap in the correction of radial club hand. J Hand Surg Br. 1995;20(3):333–7. PubMed PMID: 7561408.
28. Manske PR, McCarroll Jr HR, Swanson K. Centralization of the radial club hand: an ulnar surgical approach. J Hand Surg. 1981;6(5):423–33. PubMed PMID: 7276473.
29. Starr D. Congenital absence of the radius. A method of surgical correction. J Bone Joint Surg. 1945;27A:572–7.
30. Watson HK, Beebe RD, Cruz NI. A centralization procedure for radial clubhand. J Hand Surg. 1984;9(4):541–7. PubMed PMID: 6747239.
31. Buck-Gramcko D. Radialization as a new treatment for radial club hand. J Hand Surg. 1985;10(6 Pt 2):964–8. PubMed PMID: 4078287.
32. Kanojia RK, Sharma N, Kapoor SK. Preliminary soft tissue distraction using external fixator in radial club hand. J Hand Surg Eur Vol. 2008;33(5):622–7. PubMed PMID: 18977832.
33. Sabharwal S, Finuoli AL, Ghobadi F. Pre-centralization soft tissue distraction for Bayne type IV congenital radial deficiency in children. J Pediatr Orthop. 2005;25(3):377–81. PubMed PMID: 15832159.
34. Vilkki SK. Distraction and microvascular epiphysis transfer for radial club hand. J Hand Surg Br. 1998;23(4):445–52. PubMed PMID: 9726542.
35. Damore E, Kozin SH, Thoder JJ, Porter S. The recurrence of deformity after surgical centralization for radial clubhand. J Hand Surg. 2000;25(4):745–51. PubMed PMID: 10913218.
36. Mo JH, Manske PR. Surgical treatment of type 0 radial longitudinal deficiency. J Hand Surg. 2004;29(6):1002–9. doi:10.1016/j.jhsa.2004.06.010. Epub 2004/12/04. PubMed PMID: 15576208.
37. Cheng JC. Distraction lengthening of the forearm. J Hand Surg Br. 1991;16(4):441–5. Epub 1991/11/01. PubMed PMID: 1779163.
38. Matsuno T, Ishida O, Sunagawa T, Suzuki O, Ikuta Y, Ochi M. Radius lengthening for the treatment of Bayne and Klug type II and type III radial longitudinal deficiency. J Hand Surg. 2006;31(5):822–9. PubMed PMID: 16713850.
39. Raimondo RA, Skaggs DL, Rosenwasser MP, Dick HM. Lengthening of pediatric forearm deformities using the Ilizarov technique: functional and cosmetic results. J Hand Surg. 1999;24(2):331–8. PubMed PMID: 10194019.

40. Villa A, Paley D, Catagni MA, Bell D, Cattaneo R. Lengthening of the forearm by the Ilizarov technique. Clin Orthop Relat Res. 1990;250:125–37. Epub 1990/01/01. PubMed PMID: 2293920.
41. Waters PM, Van Heest AE, Emans J. Acute forearm lengthenings. J Pediatr Orthop. 1997;17(4):444–9. Epub 1997/07/01. PubMed PMID: 9364380.
42. Pike JM, Manske PR, Steffen JA, Goldfarb CA. Ulnocarpal epiphyseal arthrodesis for recurrent deformity after centralization for radial longitudinal deficiency. J Hand Surg. 2010;35(11):1755–61. doi:10.1016/j.jhsa.2010.07.022. Epub 2010/10/12. PubMed PMID: 20932693.
43. Rayan GM. Ulnocarpal arthrodesis for recurrent radial clubhand deformity in adolescents. J Hand Surg. 1992;17(1):24–7. PubMed PMID: 1538107.
44. Taghinia AH, Al-Sheikh AA, Upton J. Preoperative soft-tissue distraction for radial longitudinal deficiency: an analysis of indications and outcomes. Plast Reconstr Surg. 2007;120(5):1305–12; discussion 13–4. PubMed PMID: 17898604.
45. Thatte MR, Mehta R. Treatment of radial dysplasia by a combination of distraction, radialisation and a bilobed flap—the results at 5-year follow-up. J Hand Surg Eur Vol. 2008;33(5):616–21. PubMed PMID: 18694912.
46. Goldfarb CA, Murtha YM, Gordon JE, Manske PR. Soft-tissue distraction with a ring external fixator before centralization for radial longitudinal deficiency. J Hand Surg. 2006;31(6):952–9. PubMed PMID: 16843155.
47. Thirkannad SM, Burgess RC. A technique for using the Ilizarov fixator for primary centralization in radial clubhand. Tech Hand Up Extrem Surg. 2008;12(2):71–8. PubMed PMID: 18528232.
48. VanHeest A, Grierson Y. Dorsal rotation flap for centralization in radial longitudinal deficiency. J Hand Surg. 2007;32(6):871–5. PubMed PMID: 17606069.
49. Lidge R. Congenital radial deficient club hand. J Bone Joint Surg. 1969;69A:1041–2.
50. Sestero AM, Van Heest A, Agel J. Ulnar growth patterns in radial longitudinal deficiency. J Hand Surg. 2006;31(6):960–7. PubMed PMID: 16843156.
51. Albee FH. Formation of radius congenitally absent: condition seven years after implantation of bone graft. Ann Surg. 1928;87(1):105–10. PubMed PMID: 17865806.
52. Bowen CV, Ethridge CP, O'Brien BM, Frykman GK, Gumley GJ. Experimental microvascular growth plate transfers. Part I—Investigation of vascularity. J Bone Joint Surg Br. 1988;70(2): 305–10.
53. Bowen CV, O'Brien BM, Gumley GJ. Experimental microvascular growth plate transfers. Part 2—Investigation of feasibility. J Bone Joint Surg Br. 1988;70(2):311–4.
54. Donski PK, O'Brien BM. Free microvascular epiphyseal transplantation: an experimental study in dogs. Br J Plast Surg. 1980;33(2):169–78. PubMed PMID: 7388206.
55. Wall LB, Ezaki M, Oishi SN. Management of congenital radial longitudinal deficiency: controversies and current concepts. Plast Reconstr Surg. 2013;132(1):122–8. doi:10.1097/PRS.0b013e318290fca5. Epub 2013/06/29. PubMed PMID: 23806915.
56. de Jong JP, Moran SL, Vilkki SK. Changing paradigms in the treatment of radial club hand: microvascular joint transfer for correction of radial deviation and preservation of long-term growth. Clin Orthop Surg. 2012;4(1):36–44. doi:10.4055/cios.2012.4.1.36. Epub 2012/03/02. PubMed PMID: 22379554; PubMed Central PMCID: PMC3288493.
57. Horii E, Nakamura R, Nakao E, Kato H, Yajima H. Distraction lengthening of the forearm for congenital and developmental problems. J Hand Surg Br. 2000;25(1):15–21. PubMed PMID: 10763716.
58. Peterson BM, McCarroll Jr HR, James MA. Distraction lengthening of the ulna in children with radial longitudinal deficiency. J Hand Surg. 2007;32(9):1402–7. PubMed PMID: 17996775.
59. Pickford MA, Scheker LR. Distraction lengthening of the ulna in radial club hand using the Ilizarov technique. J Hand Surg Br. 1998;23(2):186–91. PubMed PMID: 9607657.
60. Geck MJ, Dorey F, Lawrence JF, Johnson MK. Congenital radius deficiency: radiographic outcome and survivorship analysis. J Hand Surg. 1999;24(6):1132–44. PubMed PMID: 10584933.
61. Dana C, Auregan JC, Salon A, Guero S, Glorion C, Pannier S. Recurrence of radial bowing after soft tissue distraction and subsequent radialization for radial longitudinal deficiency. J Hand Surg. 2012;37(10):2082–7. doi:10.1016/j.jhsa.2012.07.018. Epub 2012/10/02. PubMed PMID: 23021174.
62. Vilkki SK. Vascularized metatarsophalangeal joint transfer for radial hypoplasia. Semin Plast Surg. 2008;22(3):195–212. doi:10.1055/s-2008-1081403. Epub 2008/08/01. PubMed PMID: 20567714; PubMed Central PMCID: PMC2884879.
63. Buffart LM, Roebroeck ME, Janssen WG, Hoekstra A, Selles RW, Hovius SE, et al. Hand function and activity performance of children with longitudinal radial deficiency. J Bone Joint Surg. 2008;90(11):2408–15. PubMed PMID: 18978409.