



# Congenital Radial Dysplasia

## *Displasia radial congénita*

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### Abstract

#### Keywords

- radial dysplasia
- congenital upper limb anomalies
- congenital hand surgery

Radial dysplasia (RD) is a congenital condition which can affect the entire upper limb, compromising multiple tissues, and it always features alterations in the thumb. It can be uni- or bilateral condition, the latter being very disabling. Treatment with manipulations, casts and/or orthoses is recommended for the least severe presentations of this disease. The surgical treatment is reserved for the most severe cases, and carpal centralization is the treatment of choice today. The most frequent complication of this intervention is the recurrence of the deformity. The patients submitted to the surgical treatment report a significant improvement in the cosmetic aspect of the extremity. The current challenge is to improve the follow-up and the use of functional assessment tools regarding the results.

### Resumen

#### Palabras clave

- displasia radial
- anomalías congénitas de las extremidades superiores
- cirugía de la mano congénita

La displasia radial (DR) es una condición congénita que puede afectar toda la extremidad superior, comprometiendo diversos tejidos, y está siempre acompañada de una alteración del pulgar. La RD puede ser uni o bilateral, siendo en este último caso muy discapacitante. El tratamiento con manipulaciones, yeso y/u órtesis se indica para presentaciones leves de la enfermedad. El tratamiento quirúrgico queda reservado para los casos más severos, siendo la centralización del carpo el tratamiento de elección hoy. La complicación más frecuente de esta intervención es la recurrencia de la deformidad. Los pacientes operados mejoran significativamente el aspecto estético de la extremidad. El desafío actual es mejorar el seguimiento y el uso de herramientas de evaluación funcional respecto a los resultados.

### Introduction

Radial dysplasia (RD) is a congenital condition that encompasses a wide variety of abnormalities in the preaxial border of the forearm, especially a visible radial alteration

associated with hand deformities. It results in bone, joint, neurological, tendinous, and vascular compromise. The first author to describe this congenital pathology was Petit<sup>1</sup> in 1733.

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Epidemiologically, RD occurs in 1/50 thousand to 1/100 thousand live births,<sup>2</sup> and is slightly more frequent in males.<sup>3</sup> It always encompasses thumb abnormalities, most frequently an absent (50%), rudimentary (30%), or hypoplastic (20%) thumb, as described in different series.<sup>4,5</sup> Some cases also affect the index finger.

Although RD is a preaxial lesion, ulnar involvement is frequent. The ulna reaches only 60% of its length in moderate and severe RD.<sup>5</sup> The condition may also cause other upper extremity alterations, such as a short humerus, proximal radioulnar joint synostosis, dislocation of the radial dome, and finger stiffness.<sup>5</sup>

The unilateral presentation is the most common (60%), affecting mainly the right side.<sup>5</sup> Bilateral involvement results in considerable functional changes because the lack of a thumb associated with a radialized wrist, little movement, and a short forearm cause significant difficulties to use the hand for feeding and hygiene.

Etiology

The origin of this condition is unclear; it is known as a form of dysplasia occurring in the apical area of the ectoderm of the limb during the first weeks of embryonic development. However, the genetic bases to explain RD are limited, and its inheritance is extraordinary.<sup>3</sup>

Radial dysplasia frequently associates with severe congenital syndromes (►Table 1). As such, in the presence of unilateral or bilateral RD, a thorough examination by pediatricians and geneticists must be performed.<sup>6</sup>

Among the wide range of associated syndromes, blood dyscrasias, including Falconi anemia (with no thumb) and thrombocytopenia with absent radius (TAR) syndrome (with a thumb), are worth mentioning.<sup>6</sup>

Table 1 Syndromes associated with radial dysplasia

| Syndromes associated with radial dysplasia  |
|---|
| <i>Syndromes with blood discrasias:</i><br>Falconi anemia;<br>TAR syndrome; and<br>Aase syndrome.   |
| <i>Syndromes with cardiac abnormalities:</i><br>Holt-Oram syndrome; and<br>Lewis syndrome.  |
| <i>Syndromes with craniofacial abnormalities:</i><br>Nager craniofacial dysostosis;<br>Juberg-Hyward syndrome;<br>Baller-Gerold syndrome; and<br>Levy-Hollister syndrome. |
| <i>Syndromes with congenital scoliosis:</i><br>VATER syndrome;<br>Goldenhar syndrome; and<br>Klippel-Feil syndrome.   |
| <i>Radial dysplasia with chromosomal aberrations:</i><br>Trisomy 18;<br>trisomy 21; and<br>trisomy 13.  |

Abbreviations: TAR, thrombocytopenia with absent radius; VATER, vertebrae, anus, tracheoesophageal fistula, renal abnormalities.

Another remarkable aspect is the association between RD and cardiac abnormalities (with a rate ranging from 10% to 30%). There is a strong relationship between abnormalities of the forearm and the ventricular septum. The development of the diaphysis of the radius and humerus occurs in the fifth week of embryonic development, coinciding with the development of the ventricular septum.<sup>7</sup>

Radial Dysplasia-Related Anomalies

Flatt<sup>8</sup> published an elegant study about RD-related anatomical variations.<sup>8</sup>

i. Bones and joints

The capitate, hamate, triquetrum, and four ulnar metacarpal bones (with their phalanges) are consistently normal in virtually 100% of the cases.<sup>8</sup> The humerus is shorter and may present distal epiphyseal abnormalities (of the epitrochlea, epicondyle, olecranon fossa).<sup>8</sup>

The ulna is often curved, thickened, and shortened; the olecranon may be hypoplastic. The distal ulna has a poor growth potential compared to that of a normal limb, reaching a maximum of 60% in length. This poor ulnar growth has surgical implications because the delayed treatment has a lower chance of affecting bone development.<sup>8</sup>

Wrist anatomy is rarely unaffected. The joint capsule consists of fibrous connections alone, and mobility reaches 45° in flexion-extension.<sup>8</sup> The fingers present a characteristic stiffness in the interphalangeal joints, more accentuated in the index and middle digits.<sup>8</sup> The radius, scaphoid, trapezium, and the first metacarpal bone (with its phalanges) are absent in more than half of the patients with RD. The radius may not be entirely absent, presenting only its proximal portion, with variable length.<sup>8</sup>

ii. Muscles

The musculature of the arm is little compromised. The pectoralis major may fuse with the deltoid. The long head of the biceps is mostly absent. The short head of the biceps is always present, and may be normal. Its distal attachment is aberrant. In the forearm, the musculature originating from the epitrochlea (postaxial) is well defined proximally and may have aberrant attachments distally. The proximal attachment of the flexor carpi ulnaris is unaffected. The deep flexors of the fingers are compromised more frequently than the superficial ones, and the flexors of the index are almost always absent. The epicondyle (preaxial) musculature is more severely compromised, showing a range of variations. The extensor digitorum communis is virtually always present but fused with other extensor muscles. On the hand, the hypothenar musculature is always present.<sup>8</sup>

iii. Nerves

Radial dysplasia also involves nerves, potentially reaching down to the level of the cords in the brachial plexus. The axillary and ulnar nerves are always present and normal. The musculocutaneous nerve usually is absent, and the radial nerve disappears after innervating the triceps. The

median nerve is always present, but its distribution varies depending on the status of other nerves. Its trajectory has an invariably altered course which must be considered during the surgical treatment.<sup>8</sup>

#### iv. Blood vessels

In addition, RD is related to vascular abnormalities. The brachial and ulnar arteries are present and unaffected. However, the ulnar artery can present different divisions and projections in the attempt to supply the territory of the radial artery. The trunks of the interosseous arteries are present and well developed. It is critical to recognize the ulnar artery during surgery and dissection because the posterior interosseous artery is at risk during carpal translocation.<sup>8</sup>

## Radial Dysplasia Classification

The most accepted RD classification was devised by Heikel and modified by Bayne and Klug<sup>9</sup> (►Figure 1).

- Type I: short distal radius. The distal radial epiphysis is present but appears later. The reduced distal radius growth results in a short radius with virtually no radial deviation of the carpus. Thumb hypoplasia is frequent.
- Type II: both radial epiphyses are present but have a growth deficit, resulting in a small radius and curved ulna. The carpus lacks support and presents a radial deviation.
- Type III: partial absence of the radius. The middle and/or distal thirds of the radius are absent. The hand presents a radial displacement. The ulna is thick, short, and curved. The carpus has no support.

- Type IV: It is the most common type of RD, which was observed in 67% of the sample in the series published by Bayne and Klug.<sup>9</sup> The carpus has no support, and the hand is severely radialized.

## Treatment

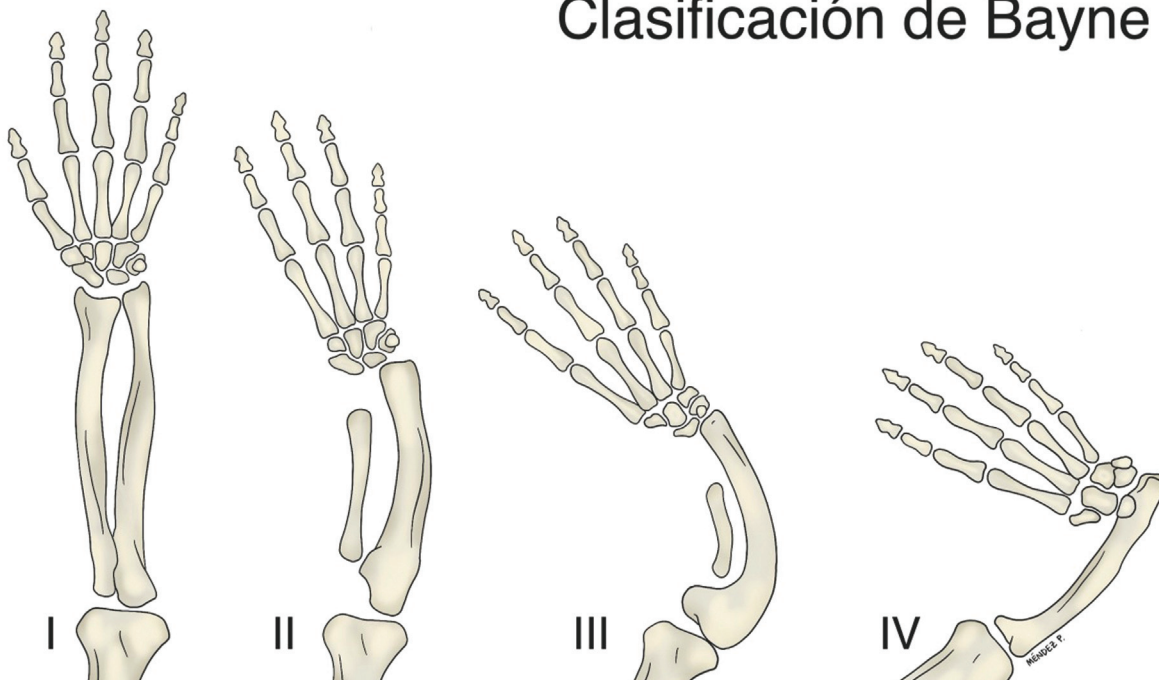
The treatment for RD is complex, requiring a balance between function and cosmetic appearance. In fact, many untreated adults manage to adapt and become self-reliant and independent in many daily-life activities, and choose activities compatible with their functional status.<sup>8</sup> Remember that any surgery can compromise function, and not altering it must remain one of the pillars of treatment.

When approaching an RD patient, keep in mind that thumb hypoplasia, wrist instability, and forearm shortness impair function.

When dealing with an abnormality in a single limb, one could take extreme measures to correct the ulna-hand axis, even risking function, due to the development of compensatory abilities during childhood. This concept is more difficult to reconcile in patients with bilateral compromise. Actions aimed at wrist stabilization will improve the performance of the extrinsic muscles of the hand, even at the expense of digital stiffness, as previously described.

In general, RD treatment has become more aggressive over the years. Today, carpal centralization, aligning the ulna with the third metacarpal, is the most accepted treatment.<sup>3,8,9</sup>

### 1. i. Manipulation and casts



**Fig. 1** Bayne and Klug<sup>9</sup> classification. Note: This drawing is owned by Magdalena Méndez Pérez. Clasificación de Bayne y Klug<sup>9</sup> = Bayne and Klug<sup>9</sup> classification



**Table 2** Contraindications for surgery in patients with radial dysplasia

| Contraindications for surgery  |
|--|
| <ul style="list-style-type: none"><li>• Children with severe abnormality, with a reduced life expectancy;</li><li>• Infants younger than 6 months of age;</li><li>• Children with severe contracture, elbow stiffness; and</li><li>• Adults with acquired abilities.</li></ul> |

The management of RD should begin as soon as possible after birth; manipulations and casts are the first lines of treatment.

Wrist and elbow manipulations may be the only treatment for type-I and some type-II RDs, and the sooner they start, the better the outcome.<sup>10</sup>

Elbow stiffness frequently responds to mobilizations, which should be short, gentle, and repeated several times each day. These actions must be well controlled to avoid fractures. Position the wrist and hand to mobilize the elbow, ideally reaching 90° of flexion.<sup>10</sup>

An alternative is to use serial plaster casts, orthoses, or both. Take great care because it is not easy to mold on such small limbs without creating compressions. The cast must include the elbow and fingers, leaving the thumb out. Ideally, change it every two weeks.<sup>10</sup>

Type-III or -IV RDs require manipulations and casts before surgery. Along with orthoses, these manipulations are effective until the patient is 2 or 3 years old.

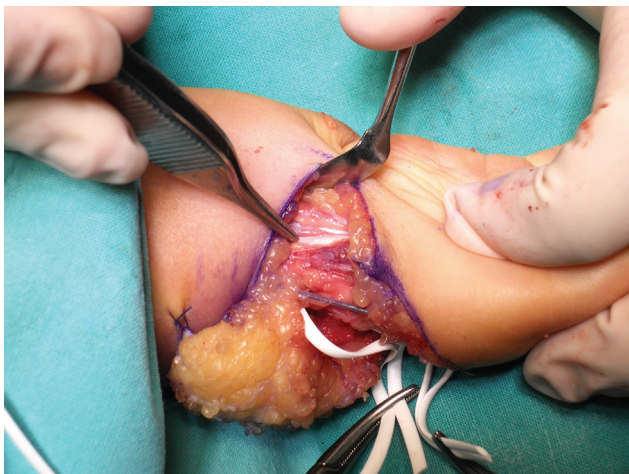
1. ii. Surgery

Carpal centralization over the distal ulna is the current mainstay of treatment. However, some conditions contraindicate this intervention (►Table 2).

The surgery starts by designing a flap enabling the transfer of good-quality skin and tissue from the ulnar side (where there is excess tissue) to the radial side (where there is a lack of tissue). Our team uses the bilobed flap proposed by Evans et al.<sup>11</sup> (►Figure 2) or the simple V flap, depending on the



**Fig. 2** Bilobed flap design to transfer good-quality skin, in a staggered manner, from the ulnar to the radial side.



**Fig. 3** The clamp shows that the median nerve is in a more radial position than usual.

size of the defect. During flap dissection, respect the venous network, access a fibrous tissue that stands out on the radial side, and section it. It is critical to identify any anchorage, or precordium, uniting the hypoplastic radius with the carpus. The presence of a precordium results in early ulnar radialization and curvature; therefore, if there is an outline of the proximal radius, search for the precordium, dissect it, and resect it.

Continue the dissection of the median nerve to identify and isolate it. This nerve is below the sectioned fibrous tissue, in a more radial position than usual (►Figure 3). Dissect the extensor apparatus of the long fingers, which is thin but always present. Mark and isolate it to protect it from damage (►Figure 4).

Next, expose the carpal capsule. Section it and carefully expose the carpal bones. This task is not easy because the carpus is in an aberrant position, and it is confused with capsular tissue. After identifying the carpus, continue with the dissection and section of the entire capsule, dorsally and palmarly, to leave the bone free of any structure.



**Fig. 4** Image of the extensor apparatus of the hand; the clamp supports the extensor apparatus with an elastic, and the other elastic supports the extensor carpi ulnaris. From this site, the radial extensors can be transferred if present.

Now, try to center the carpus on the ulna. This maneuver frequently requires making a cavity in the cartilage of the distal ulna. It is possible to do the same in the carpus. Next, stabilize the carpus by passing a Kirschner wire retrogradely. Pass the wire from the carpus to the second or third metacarpal bones. Then, introduce it into the ulna retrogradely, making sure it reaches the olecranon. This maneuver is not easy. Avoid multiple attempts of intramedullary anchorage to not cause damage to the distal ulnar epiphysis and metacarpal bones. If the ulnar curvature is significant (greater than 30°), perform an osteotomy at the most deformed site, as proposed by Goldberg and Meyn.<sup>12</sup>

With this anchorage ready, suture the preserved capsule and complete the planned transfer of the radial extensor tendons, inserting them on the ulnar side of the carpus or the extensor carpi ulnaris (ECU).

Release ischemia, check the proper vascularization of all tissues, and continue with flap overlapping and skin suture (► **Figure 5**). Place a valve, plaster, or both on the elbow, and keep it for three months. Monitor pain, finger capillary refill, and edema during the first 48 hours. After three months, place an orthosis to fix the wrist on the radial side and keep it until completing six months of treatment. Instruct the parents about the correct mobilization of the fingers and elbow and allow the child to perform grasping activities.

Surgical RD treatment must occur before the age of 3 years.<sup>8</sup> In older patients, the curved ulna may cause problems during deformity correction. The ulna virtually never curves in patients operated on at around the age of 1 year and submitted to an ulnar osteotomy.<sup>3</sup>

Another potential intervention is using external fixators to support soft tissue distraction. Some groups<sup>13,14</sup> recommend this strategy when the radial deviation of the carpus is too accentuated, and its correction is expected to be difficult. An external fixator is placed for soft tissue distraction before surgery. These devices may be circular or monoplanar fixators.<sup>13,14</sup>

Ulna lengthening is also part of the treatment. It is preferred in older children and adolescents, increasing ulnar

length up to 30%. This treatment is long and requires emotional stability and family commitment.<sup>2,15,16</sup>

In 2001, Vilkki<sup>17</sup> proposed another option to correct carpal positioning and prevent the recurrence of radial deviation. His technique uses a vascularized transfer of the second metatarsophalangeal joint.<sup>17</sup> This transfer joins the ulna, providing bony support to avoid the radial deviation of the carpus. Although this technique seems a good option, the only other article on it on the literature has been published by the same author.<sup>18</sup>

## Results

The main issue is to reproduce the radial deviation, which explains why the surgical treatment classically concentrates on measuring angles in radiographs and wrist movements.<sup>19</sup> These measurements occur in the anteroposterior and lateral planes per the criteria established by Bora et al.<sup>20</sup> Some series also include finger mobility at the end of treatment.<sup>21</sup>

In a series on centralization with tendon transfer and ten years of follow-up, Bora et al.<sup>20</sup> achieved a forearm-hand angle of 35°. In 1.5 years of follow-up, Saini et al.<sup>5</sup> achieved forearm-hand angles of 10° in the anteroposterior plane and of 6° in the lateral plane. Flatt<sup>8</sup> is aware that recurrence of the radial deviation is the major problem; therefore, he does not want a perfect outcome, and recommends centralization because the function gained is not lost.

The deceleration of the distal ulnar physis is another potential issue when the ulna impacts the carpus; sometimes, it seems to be the cost of a more functional hand. Saini et al.<sup>5</sup> showed that the length of the ulna changed from 67% to 57% in relation to the humerus, but the length of the limb remained the same. After wrist stabilization, the ranges of motion of the fingers tend to decrease a little.<sup>21</sup>

## Functional Implications

Left untreated, RD, especially when bilateral, results in severe limitations in activities such as personal hygiene, eating, and getting dressed. Carpal centering can provide a stable wrist and better grip strength. In addition, a limb with better alignment increases the ability to reach and manipulate objects. The tripod clamp is more developed in patients with a thumb. In some cases, the ulnar fingers present higher mobility and can form a slender clamp. The improved cosmetic appearance results in a positive body image on the part of the patient, increasing their social participation. However, the implementation of measurement tools to evaluate the functional outcomes of these interventions has not increased.

## Conclusion

Radial dysplasia is a congenital condition potentially affecting the entire limb and many anatomical structures. When bilateral, it is a highly-disabling condition. It is also frequently associated with syndromes, and the treatment of these children must consider the presence of other significant abnormalities. Surgical intervention, although widely



**Fig. 5** At the end of the surgery, a Kirschner wire stabilizes the wrist, and the skin closure shows the transferred flaps.

promoted, is not an easy decision because of the challenge of not sacrificing the function of these extremities. Bearing in mind that there is a significant improvement in the appearance of the limb, we still do not have sufficient long-term objective or functional measurements for our interventions. Our surgical techniques have improved, but outcome evaluation poses a significant challenge.

#### Conflict of Interests

The authors have no conflict of interests to declare.

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