

Congenital Longitudinal Radial Deficiency in Infants: Spectrum of Isolated Cases to VACTERL Syndrome

Saifullah Khalid, Mohd Faizan¹, Md Mahfooz Alam², Farogh Hassan², Samreen Zaheer, Mohd Khalid
 Departments of Radiodiagnosis, ¹Orthopedics, and ²Pediatrics, Jawaharlal Nehru Medical College and Hospital, AMU, Aligarh, Uttar Pradesh, India

ABSTRACT

Congenital longitudinal radial deficiency is a rare congenital anomaly and encompasses a spectrum ranging from mild hypoplasia to complete absence of radius. Furthermore known as radial club hand or radial dysplasia, there is variable degree of deficiency along the radial side of the limb. The authors report a case series of four cases; two cases of isolated radial club hand and two associated with other anomalies, including VACTERL syndrome. The rarity of the disease and the need to exclude other associated anomalies are emphasized.

Key words:

Congenital longitudinal radial deficiency, infants, limb anomaly, radial club hand, VACTERL syndrome

INTRODUCTION

Congenital longitudinal radial deficiency (CLRD) also known as radial club hand is a relatively rare congenital anomaly characterized by variable degree of deficiency along the radial (or preaxial) side of the limb.^[1] The estimated incidence of CLRD is found to be 1 in 30,000 to 1 in 100,000 live births.^[2-4] CLRD usually occurs sporadically with no known cause or associated with the syndrome. The spectrum of isolated anomaly to VACTERL is elucidated.

CASE REPORTS

Case 1

This was a case report of a 1-month-old-female infant was referred from evaluation of the congenital deformity of left upper limb. The baby was born to a 22-year-old primipara mother at term and her perinatal history was unremarkable. She was born at term by normal vaginal delivery following an uneventful pregnancy. She was the second child of non-consanguineous parents and there was no family history of any congenital anomaly. Physical examination of the baby was revealed deformity of the left forearm with radial deviation at the wrist and absent thumb [Figure 1a and b]. There was no anal opening at birth and an emergency colostomy was already performed. Barium study performed later confirmed rectovaginal fistula [Figure 1c]. Echocardiography showed situs solitus with dextroposition. X-ray chest also revealed D11 hemivertebrae [Figure 1d]. Ultrasonography (USG) abdomen was normal. Based on these, she was diagnosed as VACTERL syndrome. Due to associated anomalies, passive physiotherapy had been advised to the parents in consultation with physiotherapists. The baby later developed an infection in the right lung and was treated

accordingly. She has been planned for staged repair of rectovaginal fistula followed by re-anastomosis.

Case 2

Here we report a case of a 7-month-old-female child was brought by her parents with deformity of the left forearm and absent thumb, which was present since birth [Figure 2a]. The baby was the third child in the family and there was no family history of any consanguinity or similar deformity. Her intrauterine history was unremarkable. Physical examination revealed mild deformity of the left forearm, which appeared shorter than contralateral side with radial deviation at the wrist and absent thumb. Routine blood investigations were normal. The radiographs of the forearm with hands revealed type II CLRD [Figure 2b and c]. The ulna on the affected side was shorter than contralateral side. USG abdomen and echocardiography were normal. Patient was taken up for corrective surgery in the form of centralization [Figure 2d]. She is on regular follow-up and progressing well and had been planned for thumb reconstruction surgery at 18 months of age.

Address for correspondence:

Dr. Saifullah Khalid,
 House No. 386-A, Street No. 1, Iqra Colony,
 Aligarh - 202 002, Uttar Pradesh, India.
 E-mail: saif2k2@gmail.com

Access this article online	
Quick Response Code:	Website: www.jcnonweb.com
	DOI: 10.4103/2249-4847.123104

Case 3

A 6-month-old-male child was brought by his parents with deformity of the left forearm. He was born at term by normal vaginal delivery following an uneventful pregnancy. He was the second child of non-consanguineous parents and there was no family history of any congenital anomaly. X-ray forearm with hand was advised, which revealed type IV CLRD [Figure 3a]. X-ray chest revealed vertebral segmentation anomaly in the form of hemivertebrae and rib anomalies [Figure 3b]. USG abdomen and echocardiography of the neonate did not reveal any significant finding. The parents were advised consultation with the physiotherapist and regular follow-up. He is planned for centralization at around 6-9 months of age.

Case 4

A 1-month-old-male infant was referred from a peripheral health center for evaluation of the congenital deformity

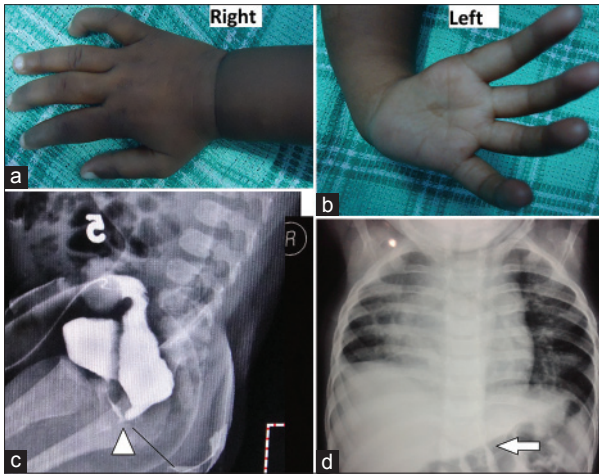


Figure 1: (a and b) 1-month-old infant with radial deviation of left forearm at wrist and absent thumb; (c) Barium study performed shows rectovaginal fistula; (d) X-ray chest also revealed D-11 hemivertebrae (bold arrow)

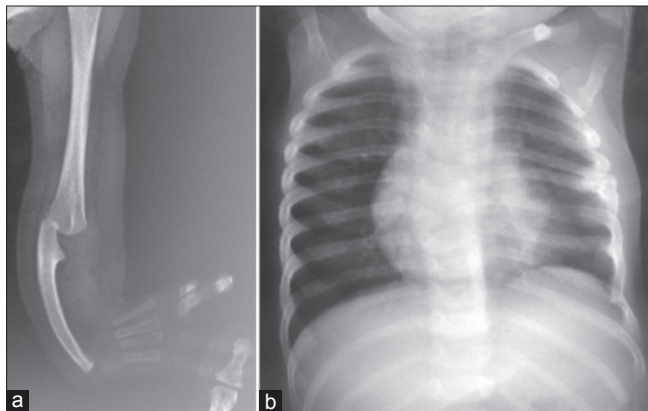


Figure 3: (a) X-ray left forearm shows type IV congenital longitudinal radial deficiency; (b) X-ray chest revealed vertebral segmentation anomaly in the form of hemivertebrae and rib anomalies

of right upper limb. The baby was born to a 25-year-old primipara mother at term and his perinatal history was unremarkable. Physical examination of the baby revealed deformity of right forearm with radial deviation at the wrist and complete absent thumb. Routine blood investigations were within normal limits. Plain radiographs of the forearm with hands showed complete absence of radius and thumb on right side [Figure 4]. USG abdomen and echocardiography of the neonate did not reveal any significant finding. The parents were advised consultation with the physiotherapist and regular follow-up. He is planned for centralization at around 6-9 months of age.

DISCUSSION

CLRD is classified by Bayne and Klug into four types. Type I is the mildest form of radial club hand. There is defective distal radial epiphysis with mild radial deviation of the

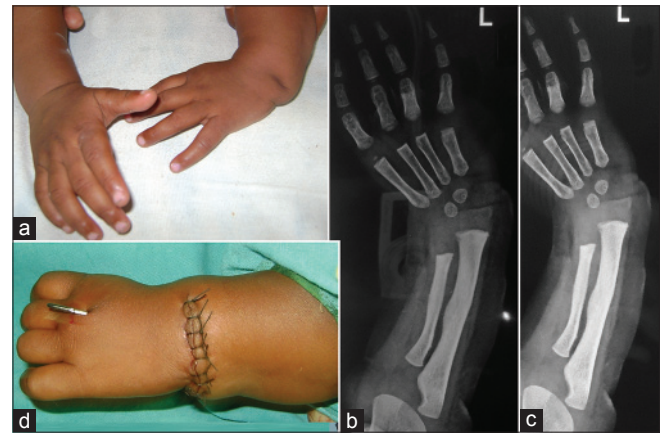


Figure 2: (a) Deformity of left forearm with radial deviation at the wrist and absent thumb; (b and c) X-ray forearm with hands revealed shortening of radius both proximally as well as distally (Type II congenital longitudinal radial deficiency); (d) Post-operative photograph after centralization procedure



Figure 4: X-ray right forearm with hand revealed complete absence of radius and thumb (type IV congenital longitudinal radial deficiency)

hand. Thumb hypoplasia can be seen. Type II shows limited growth of the radius on both the distal and proximal side. Miniature radius is seen with radial deviation of the wrist. In Type III there is partial absence of radius, most commonly of the distal two-third with severe radial deviation of the wrist. Type IV is the most common and most severe type. There is a complete absence of the radius.^[2,5]

In the present small series of four cases all the babies had unilateral involvement and no sex predilection was seen. Banskota *et al.* in his series of 19 unilateral cases had reported a 3:1 male preponderance. In his series, 8 out of 27 cases had bilateral involvement (29.6%) and no sex predilection was seen in bilateral cases. Overall, there was an increased male predilection of 2:1.^[6] Type IV CLRDR was the most common type in our series, similar to that noted by Banskota *et al.* in their series. One case of Type II was also encountered in our series, which was the least common type in their series.^[6,7]

About 40% of patients with unilateral radial club hand and 27% with bilateral radial club hand have associated congenital anomalies involving cardiac, renal, anal, skeletal and hematopoietic system.^[3] The common syndromes associated with CLRDR include Holt Oram syndrome, Thrombocytopenia absent radius syndrome, fanconi anemia and VACTERL syndrome.^[8-10] In our series, two cases of isolated CLRDR,^[11] whereas two patients had associated vertebral anomalies and one had absent anal opening with rectovaginal fistula. Oral *et al.* in their series of 28 cases of VACTERL, also reported vertebral anomalies as the most common component.^[10] None of the patients had tracheoesophageal, renal or major cardiac anomaly except for one child who had dextraposition of heart. Babies are diagnosed with VACTERL syndrome when they have at least three or more of these individual anomalies (vertebral, anal, cardiac, tracheoesophageal, renal and limb anomaly).^[10]

Treatment is guided first by any associated anomalies because most of them such as cardiac or gastrointestinal require early surgical management and are given preference.

For isolated cases management is conservative as well as surgical.^[12] Principle guiding surgical management is wrist stabilization and later thumb reconstruction.^[13]

REFERENCES

1. Goldberg MJ, Meyn M. The radial clubhand. *Orthop Clin North Am* 1976;7:341-59.
2. Bayne LG, Klug MS. Long-term review of the surgical treatment of radial deficiencies. *J Hand Surg Am* 1987;12:169-79.
3. Wahab S, Ullah E, Khan RA, Sherwani MK. Radial club hand-A case report and review of literature. *Bombay Hosp J* 2009;51:94-6.
4. Phatak SV. Radial club-hand: A case report. *Indian J Radiol Imaging* 2006;16:609-10.
5. Waters MP. The upper limb. In: Morrissy RT, Weinstein SL, editors. *Lovell and Winters Pediatric Orthopaedics*. 5th ed. Philadelphia (PA): Lippincott Williams and Wilkins; 2001. p. 841-903.
6. Banskota AK, Bijukachhe B, Rajbhandary T, Pradhan I, Singh A. Radial club hand deformity – The continuing challenges and controversies. *Kathmandu Univ Med J (KUMJ)* 2005;3:30-4.
7. Siddiqui YS, Abbas M, Khan AQ, Jilani LZ. A child with congenital longitudinal radial deficiency. *Congenital radial club hand. Saudi Med J* 2011;32:199-200.
8. Omran A, Sahnoud S, Peng J, Ashhab U, Yin F. Thrombocytopenia and absent radii (TAR) syndrome associated with bilateral congenital cataract: A case report. *J Med Case Rep* 2012;6:168.
9. De Kerviler E, Guermazi A, Zagdanski AM, Gluckman E, Frijia J. The clinical and radiological features of Fanconi's anaemia. *Clin Radiol* 2000;55:340-5.
10. Oral A, Caner I, Yigiter M, Kantarci M, Olgun H, Ceviz N, *et al.* Clinical characteristics of neonates with VACTERL association. *Pediatr Int* 2012;54:361-4.
11. Salati SA, Rabah SM. Congenital isolated right radial club hand. *Libyan J Med* 2010;5:5090.
12. Maschke SD, Seitz W, Lawton J. Radial longitudinal deficiency. *J Am Acad Orthop Surg* 2007;15:41-52.
13. Fujiwara M, Nakamura Y, Nishimatsu H, Fukamizu H. Strategic two-stage approach to radial club hand. *J Hand Microsurg* 2010;2:33-7.

How to cite this article: Khalid S, Faizan M, Alam MM, Hassan F, Zaheer S, Khalid M. Congenital longitudinal radial deficiency in infants: Spectrum of isolated cases to VACTERL syndrome. *J Clin Neonatol* 2013;2:193-5.

Source of Support: Nil, **Conflict of Interest:** None declared.