Radial Longitudinal Deficiency: The Incidence of Associated Medical and Musculoskeletal Conditions

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Purpose: Radial longitudinal deficiency (RLD) is associated with certain syndromes and medical and musculoskeletal conditions. The purpose of this investigation was to evaluate the incidence of these conditions with RLD.

Methods: A comprehensive chart review identified patients with RLD and a complete medical record. These charts were evaluated for the presence of associated medical and musculoskeletal conditions and biographic information on gestation, delivery, and family history.

Results: A total of 164 patients with 245 affected extremities were identified; 138 patients had radius abnormalities and 26 patients had isolated thumb hypoplasia. Twenty-five patients had thrombocytopenia absent radius syndrome; 22 patients had vertebral, anal, cardiac, tracheoesophageal, renal, and limb abnormalities association; 7 patients had Holt-Oram syndrome; and 1 patient had Fanconi anemia. There were 32 patients with cardiac abnormalities and 60 patients with spinal or lower-extremity musculoskeletal abnormalities. The percentage of patients with associated abnormalities increased with an increasing severity of RLD. One hundred two of the 138 patients with types I through V RLD had associated medical or musculoskeletal abnormalities. In contrast, only 9 of 26 patients with an isolated thumb hypoplasia (type 0 RLD) had associated abnormalities.

Conclusions: The high incidence of associated medical and musculoskeletal abnormalities in patients with RLD emphasizes the importance of a complete assessment including a complete musculoskeletal examination, cardiac auscultation, complete blood count, echocardiogram, renal ultrasound, and spinal radiographs. Although approximately one third of patients in this investigation had a syndrome commonly associated with RLD, most patients with RLD types I through V had an additional medical or musculoskeletal anomaly. Patients with type 0 RLD were less likely to have comorbidities. (J Hand Surg 2006;31A:1176–1182. Copyright © 2006 by the American Society for Surgery of the Hand.)

Key words: Radial longitudinal deficiency, clubhand, syndrome, cardiac.

The association of radial longitudinal deficiency (RLD) with certain medical conditions is well established; however, there are few specific data on the incidence of these various conditions. This is in part because most reports in the hand surgery literature address the classification of and surgical treatment for RLD, not the conditions associated with it. Kelikian[1] reported that 19th-century researchers gave patients with RLD a very poor general prognosis, presumably related to the associated medical conditions. A variety of medical conditions have been reported in association with RLD,[2] most commonly thrombocytopenia absent radius (TAR) syndrome[3]; the vertebral defects, anal atresia, cardiac malformation, tracheoesophageal fistula, esophageal atresia, renal anomalies, and limb anomalies (VACTERL) association[4]; Fanconi anemia (an autosomal-recessive disorder that can affect the bone marrow with associated abnormalities of the limbs, heart, and kidneys)[5]; and Holt-Oram syndrome (an autosomal-dominant disorder with RLD and heart abnormalities, typically an atrial or ventricular septal defect).[6–10] Although our understanding of these medical conditions has evolved and medical treatment has made

great progress, there is relatively little information on the relationship of these conditions to RLD.

James et al,[11] in a report on 139 patients with RLD, found that 55 patients had 1 of 12 associated syndromes; VACTERL and Holt-Oram were the most common. Flatt[2] noted that congenital heart disease occurs in 10% to 13% of patients with RLD, and Birch-Jensen[12,13] noted that more than 25% of patients had a (nonspecified) congenital heart disease. Kelikian[1] estimated that approximately 5% of patients with RLD had a blood dyscrasia. Bayne and Klug[14] reported 1 patient with Fanconi anemia and 2 patients with TAR syndrome in a series of 101 patients. Alter[15] reported 5 patients with Fanconi anemia and 6 with TAR syndrome in 129 patients with nonthalidomide radial ray dysplasias, Lamb[9] reported no associated medical conditions in 68 RLD patients, and another investigation[16] only mentioned the associated medical conditions briefly.

The current investigation was performed to determine the incidence of these medical conditions in patients with RLD presenting to the hand clinic at a large pediatric orthopedic hospital.

Materials and Methods

After institutional review board approval, we reviewed the medical records and surgical logs of our pediatric orthopedic hospital from 1949 to 2005 for patients with a diagnosis of types 0 through V RLD (Table 1).[14,11,17] In this retrospective review, multiple search terms in the medical record were used to maximize the identification of patients with RLD; these terms included *radial clubhand*, *radial longitudinal deficiency*, *hypoplastic thumb*, *upper-extremity deficiency*, *amelia*, and *phocomelia*. Patients with an incomplete medical record, with a single outpatient visit to our hospital, or without a documented thorough medical evaluation from our hospital or from their pediatrician were excluded. A total of 164 patients with RLD met these inclusion criteria and were included in this investigation.

Table 1. Extended Bayne and Klug[14] Radial Longitudinal Deficiency Classification

Type	Description
0	Hypoplastic thumb with normal radius
I	Short distal radius
II	Radius in miniature
III	Absent distal radius
IV	Complete absence of radius
V	Absent proximal humerus in addition to typical RLD abnormalities distally

Table 2. Radial Longitudinal Deficiency Classification According to Patients

Type	Unilateral	Bilateral	Total
0	14	12	26
I	8	7	15
II	6	4	10
III	5	4	9
IV	49	34	83
V	1	5	6
Mixed		15*	15*
Total	83	81	164

*Right and left extremities affected with different RLD types.

A pediatrician (either a referring pediatrician or staff pediatrician) and a hand surgeon evaluated all patients. The medical records were reviewed to identify a variety of demographic data including patient gender, race, unilateral versus bilateral involvement, type of RLD, and degree of thumb involvement. The records were specifically assessed for any associated medical or musculoskeletal diagnoses including the presence of syndromes, associations, and medical conditions. The preoperative work-up included a complete blood count and upper-extremity radiographs in all patients, but other testing was performed at the discretion of the surgeon or pediatrician. Additional testing was performed if the physical examination suggested an abnormality. All available data including echocardiogram, renal ultrasound, laboratory data, and spine evaluation results were reviewed. The physical examination was reviewed, particularly the cardiac and musculoskeletal examinations. When applicable, the preoperative anesthesia cardiac evaluation also was assessed. Echocardiograms or electrocardiograms were available for review in 31 patients. The family history was assessed for both musculoskeletal and syndrome data. The medical records also were searched for information concerning maternal risk factors during pregnancy (gestational or delivery problems).

A statistical analysis was performed using the chi-square test. Significance was set at a p value of less than .05.

Results

A total of 164 patients were identified with 245 affected extremities. Most patients were white (143); 17 patients were African American, 3 patients were Hispanic, and 1 patient was Asian. There were 90 boys and 74 girls. There were 83 patients affected unilaterally (36 right extremities, 47 left extremities),