Radial Longitudinal Deficiency

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Epidemiology

- Results from a radial ray deficiency during embryological development.
- Incidence ranges from 1 in 30,000 to 1 in 100,000.
- Ranges from 4.7% to 6.1% of all congenital anomalies.
- Slightly more common in males than females and in caucasians.
- Bilateral in 38 to 50 percent of cases.
- When unilateral, occurs twice as frequent on right side.
Radial Clubhand

- Deformity is radial deviation of hand with a short forearm (50-75% the length of normal forearm).
- Almost always present at birth.
- Prominent knob at distal end of ulna.
- Thumb may be absent or severely deficient.
- Hand typically small.
- MCP joints with limited flexion and some hyperextensibility. May be flexion contractures of PIP joints.
- Elbow extension contracture common as a result of weak or absent elbow flexors.
Etiology

- Primary insult is to the apical ectodermal ridge during critical limb development period. (between 4th and 7th weeks)
- Mostly due to environmental factors.
  - Compression
  - Inflammatory processes
  - Nutritional deficiency
  - Irradiation
  - Infection
  - Chemical exposure
  - Medications (especially thalidomide)
Etiology

- No single definite implicating factors.
- Genetic studies have failed to show any genetic basis except when deformity is associated with a syndromal picture as in Holt-Oram, Fanconi’s, and TAR syndrome (more later on these).
- Interestingly, one study found that twice as many affected patients born during summer months than winter months.
Bony Anatomy

- Scapula, clavicle, and humerus reduced in size.
- Ulna typically short, thick, and bowed. May develop synostosis with radial anlage.
- Scaphoid, trapezium absent in more than 50%.
- Lunate, trapezoid, and pisiform deficient in about 10%.
- Thumb metacarpal and its phalanges absent in more 80%, however a rudimentary thumb may be present.
- The capitate, hamate, triquetrum, and ulnar four metacarpals and phalanges present and without deficiency.
Muscle Anatomy

- Preaxial musculature from lateral epicondyle most severely affected.
- Radial wrist extensors (ECRL, ECRB and BR) either absent or severely deficient.
- Finger extensors usually present.
- Typically see abnormal insertions of FCR.
- Long head of biceps almost always absent. Short head typically hypoplastic.
- Brachialis deficient or absent
Nerve Anatomy

- Median nerve thickened and runs just below fascia. At risk for injury during surgical dissection along concavity of deformity.
- Radial nerve typically ends at lateral epicondyle after innervating triceps.
- Ulnar nerve normal.
- MC nerve absent.
Vascular Anatomy

- Radial artery absent.
- Normal brachial and ulnar arteries.
- Well-developed common interosseous artery.
Associated Deformities

- Associated anomalies frequently encountered. These can be isolated or associated with a syndrome.
- These deformities include cardiac, GI, pulmonary, genitourinary, neurologic, and skeletal malformations.
Holt-Oram Syndrome

- Autosomal dominant mode of inheritance.
- See radial deficiencies of both thumb and radius.
- May see triphalangeal thumb, radioulnar synostosis, and proximal humeral abnormalities.
- The big association with this syndrome is cardiac defects.
Holt-Oram Syndrome

- Cardiac defects most frequently seen are ASD, VSD, tetralogy of Fallot, mitral valve prolapse, PDA, total anomalous pulmonary venous return.
- Congenital heart defects required for diagnosis.
TAR Syndrome

- Characterized by Thrombocytopenia and Absent Radius.
- Thrombocytopenia begins in neonatal period.
- Thumb is always present and radial deficiency is bilateral.
- Autosomal recessive mode of inheritance.
- Typically, prognosis is good and platelet count improves to normal by age 4 to 5.
- Always check platelet count in child with Radial clubhand and a thumb prior to entertaining surgery.
Fanconi’s Anemia

- Autosomal dominant
- Thumb always present
- A progressive pancytopenia
- May not progress until mid-childhood.
- Use mitomycin C test to diagnose or confirm.
- Prognosis is poor.
VATER

- **Vertebral anomalies, Anal atresia, Tracheoesophageal fistula, Esophageal atresia, and Renal defects.**

- Some or all of these abnormalities may present at the same time as radial deficiency.

- In some of these patients, a Cardiac anomaly, Lower limb defect, and Single umbilical artery is seen. (VACTERLS)
Trisomy 13 and 18

- Radial ray deficiencies seen frequently.
- However, prognosis is typically poor secondary to severe abnormalities.
- Surgical correction usually not indicated.
Diagnostic Considerations

- Absence or presence of thumb may help in differentiating syndrome from sporadic deficiency.
- Cardiac, hematologic, radiographic, and renal workup should be ordered on all children noted to have radial ray deficiency.
Classification

- Current classification described by Heikel.
  - Type I - Short distal radius. Distal physis present but delayed in appearance. Proximal physis normal.
  - Type II – Hypoplastic radius. Distal and proximal physes present but delayed in appearance.
  - Type III – Partial absence of radius. May be proximal, middle, or distal. (Distal 1/3 absence most common)
  - Type IV – Complete absence of radius
Classification

- Heikel's classification of radial dysplasia.
  - **A,** Type I—short distal radius.
  - **B,** Type II—hypoplastic radius.
  - **C,** Type III—partial absence of radius.
  - **D,** Type IV—total absence of radius
Xray
Non-Operative Treatment

- All treatment should begin at birth and should consist of manipulation and serial splinting/casting into corrected position.
- Should be done with a well-padded long arm cast with arm gently placed into flexion and hand in maximal correctable position.
- Cast on two weeks, then off with 1 week stretching/manipulation, then new cast x 2 weeks.
Non-Operative Treatment

- Riordan advocated a staged method of correction similar to the Ponseti technique for clubfoot.
  - Hand and wrist corrected first, then elbow corrected as much as possible.
Surgical Contraindications

- Life-threatening comorbidities
- Older patients with adequate function and psychological concerns.
- Bilateral involvement with stiff elbows.
- Type I and mild type II.
Pre-op Considerations

- It has been shown by Lamb, et al. that elbow flexion does not improve after surgical correction of hand.
- Be mindful that in patients with extension contracture at the elbow, correction of the hand will render them unable to reach their mouth.
- It is imperative that elbow motion be assessed and addressed prior to surgical correction.
Operative Treatment

- In any case of operative treatment it is imperative that the carpus be able to mobilized over the end of the ulna.

- If this is not achievable after casting/splinting, lengthening through a distraction device or external fixator should be undertaken prior to formal fixation.
Operative Treatment

- Operative treatment largely involves the act of “centralizing” the carpus on the distal ulna.

- However, some feel centralization is not enough at may lead to recurrent deformity. These authors suggest “radialization” of the ulna.
Techniques

- Centralization arthroplasty technique, transverse ulnar approach.
- **A**, Incision.
- **B**, Exposure of muscle, tendon, and nerve.
- **C**, Capsular incision.
- **D**, Exposure of carpoulnar junction and excision of segment of carpal bones.
- **E**, Insertion of Kirschner wire.
- **F**, Reattachment of extensor carpi ulnaris tendon.
Techniques

- Centralization of radial clubhand.
- A, Z-plasties on radial and ulnar sides of wrist.
- B, Incisions allow lengthening on radial side. Ulnar incision takes up skin redundancy, transposing it to deficient radial side.
- C, Radial incision in wrist for identification of median nerve.
- D, View from ulnar incision across wrist to radial incision after resection of all nonessential central structures.
- E, Distal ulna seen through radial incision at wrist.
- F, Kirschner wire passed through lunate, capitate, and long finger metacarpal.
- G, After centralization, Kirschner wire passed into ulna to maintain position.
Techniques

- Centralization of hand and tendon transfer.
- **A**, Volar aspect of radial clubhand deformity showing right-angle relationship of hand and forearm and acute angulation of extrinsic flexor tendons.
- **B**, Volar aspect after centralization and transfer of sublimis tendons of ring and long fingers
Techniques

- Centralization of radial clubhand.
- **A**, Radial release and resection of redundant soft tissue.
- **B**, Centralization and pin fixation with ulnar osteotomy.
- **C**, Radial capsular release and tendon transfer.
19 radial club hands (16 type IV, 3 type III) treated with centralization followed over 6.5 years.

“Average preoperative angulation measured 83° (range, 55° to 110°). Centralization corrected the angulation an average of 58° (range, 15° to 95°) to an average immediate postoperative total angulation of 25° (range, 5° to 60°). At the final follow-up examination there was a loss of 38° (range, 5° to 105°) and the total angulation increased to an average of 63° (range, 20° to 120°). The difference between the preoperative, postoperative, and follow-up angles was statistically significant.”
Techniques


- Improve the ulnar lever arm by creating a more ulnar deviation and transferring radial extensor and flexor tendons to the ulnar side of the carpus.

- This is thought to reverse imbalance of tight radial muscles and provide a better mechanical advantage.
29 limbs in 23 patients with an average follow-up period of 50 months treated with radialization or centralization and compared outcomes.

Radialization was similar to modified centralization in the final outcome. Survivorship analysis was performed using revision as the end point, with a survivorship rate at 5 years of 67%.

“These data offer support for the hypothesis that a more ulnar translation and an ulnar angulation of the wrist is a means of reducing the radial lever arm and thus the incidence of deformity recurrence and need for revision.”
Summary

- Always be mindful of associated syndromal patterns and concomitant medical problems when approaching radial clubhand deformity.
- Set reasonable treatment goals and counsel families on reasonable expectations.
- Know that elbow flexion typically does not improve after surgery, so it is imperative to address this pre-op in order to maintain ADLs.
Bibliography

Bibliography

- Johnson, Thomas R: **Essentials of Musculoskeletal Imaging.** Copyright © 2004 AAOS.