Congenital Radioulnar Synostosis

Contents

Although congenital proximal radioulnar synostosis is an uncommon malformation of the upper extremity, first described by Sandifort in 1973 (Simmons et al. 1983), congenital radioulnar synostosis (RUS) is the most common congenital functional disorder of the elbow joint (Siemianowicz et al. 2010). In most cases, RUS is an isolated defect involving only one limb. However, bilateral RUS is usually familial and affects males more than females (Figs. 1–3).

Synonyms and Related Disorders

Proximal radioulnar synostosis

Genetics/Basic Defects

1. Etiology (Rizzo et al. 1997; Elliott et al. 2010; Wurapa 2012)
    1. Isolated or congenital radioulnar synostosis types I and II (Davenport et al. 1924)
    2. Familial cases: inherited as autosomal dominant trait (Hansen and Andersen 1970; Spritz 1978; Rizzo et al. 1997)
    3. Association of RUS and chromosome anomalies: well known (Rizzo et al. 1997; Kidszun et al. 2012)
        1. X chromosome anomalies: polysomy X in both male and female patients with supernumerary X chromosomes (49,XXXXY, 49,XXXXX, 48, XXXX, 48,XXXY, 48,XXYY, 47, XXY)
        2. Y chromosome anomalies (De Smet and Fryns 2008; Syed and Quinton 2008)
        3. Other chromosome anomalies:
            1. Del(10)(pter-p13)
            2. Del(11)(q23)
            3. Del(13)(q22-qter)
            4. Dup(12)(q24-qter)
            5. Dup(14)(q23-qter)
            6. Partial trisomy 11q
            7. Trisomy 8 mosaicism
            8. Trisomy 18p
    4. Association of RUS with various syndromes
        1. Apert syndrome
        2. Arthrogryposis
        3. Berant syndrome (radioulnar synostosis and craniosynostosis)
        4. Carpenter syndrome

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5. Cenani-Lenz syndactyly: a form of syndactyly resembling that of Apert syndrome but with additional features such as severe shortening of the ulna and radius with fusion, fusion of the metacarpals, "disorganization" of phalangeal development, and less severely affected feet (Cenani and Lenz 1967)
6. De Lange syndrome
7. Der Kaloustian syndrome (radioulnar synostosis, short stature, retinal changes)
8. Fetal alcohol syndrome (Froster and Baird 1992)
9. Fetal thalidomide syndrome (Smithells and Newman 1992)
10. Fetal vitamin A syndrome (Rizzo et al. 1991)
11. Holt-Oram syndrome
12. Jorgenson syndrome (blepharophimosis and radioulnar synostosis)
13. Mandibular dysostoses
14. Michels syndrome (clefting, ocular anomalies, radioulnar synostosis)
15. Multiple dysostosis
16. Nail-patella syndrome
17. Noonan syndrome
18. Poland syndrome
19. Williams syndrome
20. Others

5. Developmental gene(s) responsible has not been identified in isolated RUS (Elliott et al. 2010)
    1. Mutations in the homeobox A11 (*HOXA11*) gene resulting in autosomal dominant RUS with megakaryocytic thrombocytopenia in two pedigrees have been reported (Thompson and Nguyen 2000)
    2. The HOX genes: critical developmental genes in limb development
    3. Not all patients with the gene mutation demonstrated marrow failure

2. Developmental basis of proximal radioulnar synostosis (Elliott et al. 2010)
    1. The upper limb bud arises from the body wall at approximately 26 days of development (after conception)
    2. The elbow is first discernible at 35 days: presence of three connected cartilaginous anlagen which will eventually develop into the humerus, radius, and ulna
    3. Soon afterward, longitudinal segmentation produces separation of the distal radius and ulna. However, briefly, their proximal ends are united and share a common perichondrium (Lewis 1901)
    4. Abnormal genetic or environmental factors operating at this time in development could interrupt subsequent proximal radioulnar joint morphogenesis (Mital 1976)
    5. Such interference would allow for later ossification of the entire proximal cartilaginous model and produce complete bony synostosis. If joint development continued before the developmental arrest occurred, this could lead to a smaller area of coalition and the presence of a rudimentary radial head (Simmons et al. 1983)
    6. These two scenarios also represent related primary anomalies of radioulnar differentiation/segmentation. The final specific defect of this spectrum is influenced by subtle differences in developmental timing

Clinical Features

1. 2 types of radioulnar synostosis (Wilkie 1914; Bauer and Jonsson 1988)
    1. Type 1
        1. Proximal smooth fusion of 2–6 cm between the radius and ulna
        2. Absent radial head
        3. Resulting in a limitation of pronation and supination of the forearm
    2. Type 2
        1. A fusion just distal to the proximal radial epiphysis

   2. Association with congenital dislocation of the radial head
   3. Resulting in a limitation of pronation and supination of the forearm and a restriction of extension at the elbow
2. Pain
   1. Not always associated with radioulnar synostosis
   2. Occurrence of pain: usually thought to be due to progressive, symptomatic radial head subluxation (Sachar et al. 1994)

Diagnostic Investigations

1. Radiographic types (Cleary and Omer 1985)
   1. Type I (19%): fibrous synostosis with a reduced normal-appearing radial head
   2. Type II (8%): visible bony synostosis with a reduced radial head
   3. Type III (56%): visible bony synostosis with a hypoplastic and posteriorly dislocated radial head
   4. Type IV (17%): short bony synostosis with an anteriorly dislocated mushroom-shaped radial head
2. CT scans recommended especially in cases in which plain radiographs show no osseous fusion (Karatosun et al. 2004)
3. MRI: demonstrates soft tissue anatomy better

Genetic Counseling

1. Recurrence risk
   1. Patient's sib
      1. Sporadic: probably not increased
      2. Autosomal dominant inheritance: not increased unless a parent is affected
   2. Patient's offspring
      1. Sporadic: probably not increased
      2. Autosomal dominant inheritance: 50%
2. Prenatal diagnosis of bilateral radioulnar synostosis by ultrasonography in a fetus of 49, XXXXY female at the second trimester of gestation (Martini et al. 1993)
3. Management
   1. Patients with congenital radioulnar synostosis in nearly neutral rotation could perform activities of daily living with the aid of compensatory movements of the shoulder and elbow (Kasten et al. 2009).
   2. Surgical intervention (Mostert and Tulp 2002):
      1. Usually not recommended, except in bilateral cases or in hyperpronation
      2. Optimal age for surgery difficult to define
         1. 4–5 years of age in cases of hyperpronated hand
         2. 12 years of age in the case of bilateral synostosis
   3. Derotational osteotomy at the shafts of the radius and ulna for congenital radioulnar synostosis (Murase et al. 2003).
   4. Surgical treatment with radial head excision yielded resolution of pain and restoration of elbow flexion (VanHeest et al. 2013).

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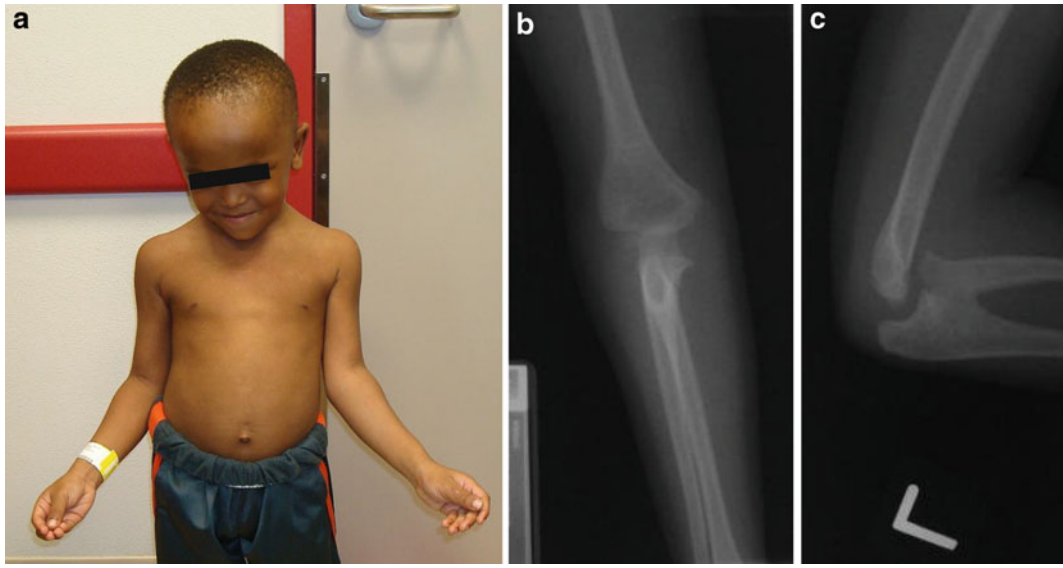

Fig. 1 (**a**–**c**) This 3½-year-old boy was evaluated for familial bilateral proximal radioulnar synostosis (shown by radiograph) (**b**, **c**). He had a large head, frontal bossing, and depressed nasal bridge is and unable to supinate both wrists (**a**)

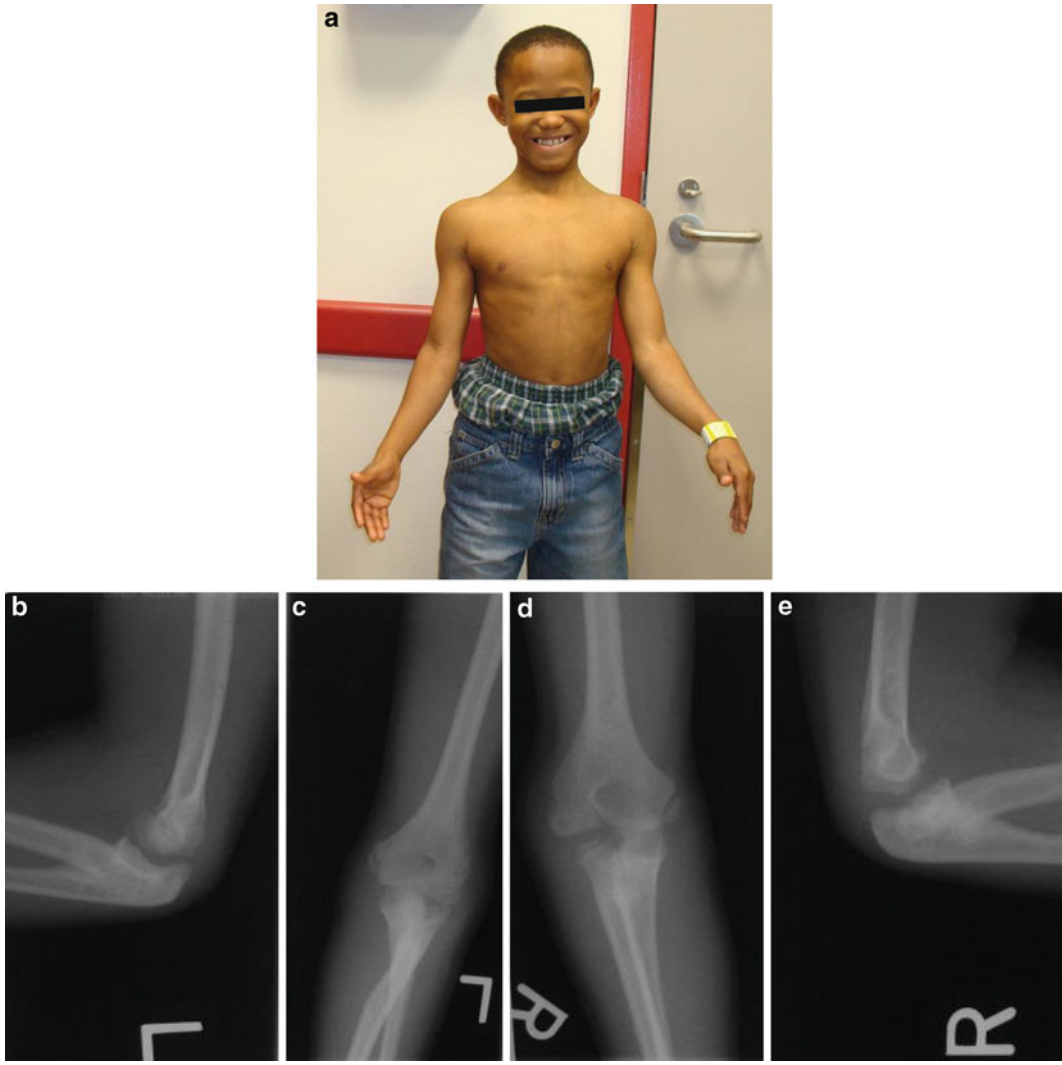

Fig. 2 (**a**–**e**) The 10-year-old brother also had bilateral proximal radioulnar synostosis, shown by radiographs (**b**–**e**) with similar clinical features (**a**)

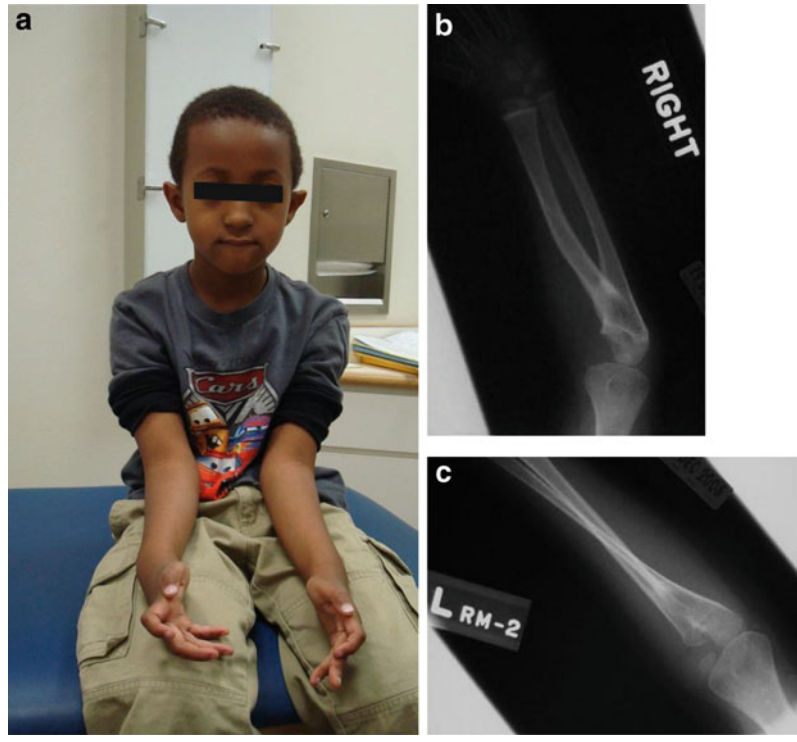

Fig. 3 (**a**–**c**) This young boy was evaluated because of bilateral cubitus valgus (**a**). He could not extend his forearms fully and supinate his wrists. Radiographs showed bilateral proximal radioulnar synostosis (**b**, **c**)