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Case Report

Sporadic Class II Congenital humeroradial synostosis and Left Micromelia in a three-and-a-half-months female Ghanaian infant☆,☆☆

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ABSTRACT

Congenital humeroradial synostosis (CHRS) is a rare musculoskeletal condition that significantly affects the mobility of the elbow joint. They occur in various types and forms depending on the types and numbers of bones involved at the elbow. CHRS may present with elbow deformity and limitation of function. Appropriate timely diagnosis and counseling are required since CHRS is mostly managed conservatively according to literature and may prevent avoidable fractures of the radius from attempts by parents to straighten the flexed fixed elbow and finally offer adequate time for delayed surgical intervention which is usually ineffective and unhelpful.

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Introduction

Congenital humeroradial synostosis (CHRS) is 1 of 3 rare developmental anomalies occurring in the elbow joint, with a reported prevalence of < 1/1,000,000, the other 2 being fusions between the humerus and ulna, or with both radius and ulna (with humeroradial synostosis being the commonest among the elbow joint bone fusions) [1,2]. Radioulnar synostosis though also rare, is more common than fusion abnormalities about the elbow joint [3]. The exact cause of CHRS is unknown however, the condition has been linked to genetic factors and cocaine use during pregnancy [4].

Genetic or familial cases are believed to be associated with syndromes which may involve abnormalities such as microcephaly, short corpus callosum, and ambiguous genitalia [5–7]. Sporadic cases have also been reported and are believed to be associated with upper limb hypoplasia [5].

Undiagnosed cases of CHRS may present with fixed elbow joint in flexion in infants, which may be complicated by radial bone fractures with repeated attempts by parents to straighten the flexed joint [5]. Appropriate timely diagnosis and counseling are required since CHRS is mostly managed conservatively according to the literature [5,7]. Unlike the many reports of radioulnar synostosis from Africa and other continents, our search of the literature has yielded no report of humeroradial synostosis from West Africa [8–10]. We hereby present a case of unilateral humeroradial synostosis that was diagnosed at our imaging facility in Accra, Ghana.

Case report

A 3-and-a-half-month-old female infant was seen for radiographs of both elbow joints at our X-ray unit, on account of a left upper limb abnormality since birth. The radiographs demonstrated a fusion of the left proximal radius and distal humerus. The left thumb and first metacarpal were absent. The right upper limb was normal in appearance Fig. 1.

The baby was presented to the orthopedic unit of the Korle-Bu Teaching Hospital at 3 months of age by its mother after noticing the absence of the left thumb at birth and the infant's inability to move the left elbow joint.

The pregnancy had been uneventful and the mother denied suffering any illness during pregnancy. She however admitted to consuming about 16 units of alcohol per week and being exposed to second-hand cigarette smoke at work till 6 weeks into her pregnancy when she stopped working, she however had a smooth spontaneous vaginal delivery at term. There was no family history of musculoskeletal abnormalities from both parents.

Physical examination demonstrated a stiff left elbow joint, flexed at about 80^0, and supple left shoulder and wrist joints. Both wrist joints were not deviated. The left forearm was 2 cm shorter than the right. The baby had no syndromic facial features and exhibited no other systemic abnormalities.

The left humerus, radius, and ulna were shorter (measuring 6.6 cm, 4.5 cm and 5.1 cm respectively) than the contralateral humerus, radius, and ulna (measuring 7.6 cm, 6.1 cm, and

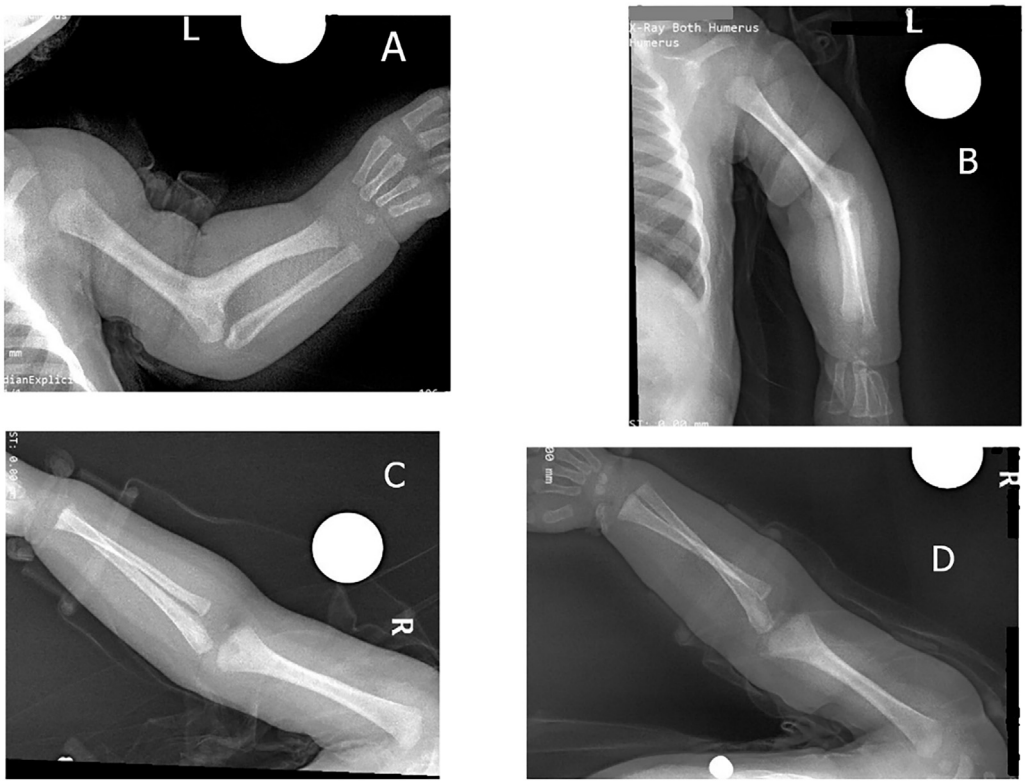

Fig. 1 – Radiographs of both upper limbs. Images A and B are of the left upper limb and show a fusion of the distal humerus and proximal radius, absence of the first metacarpal, and the first proximal phalanx. Images C and D show the normal appearance of the right upper limb.

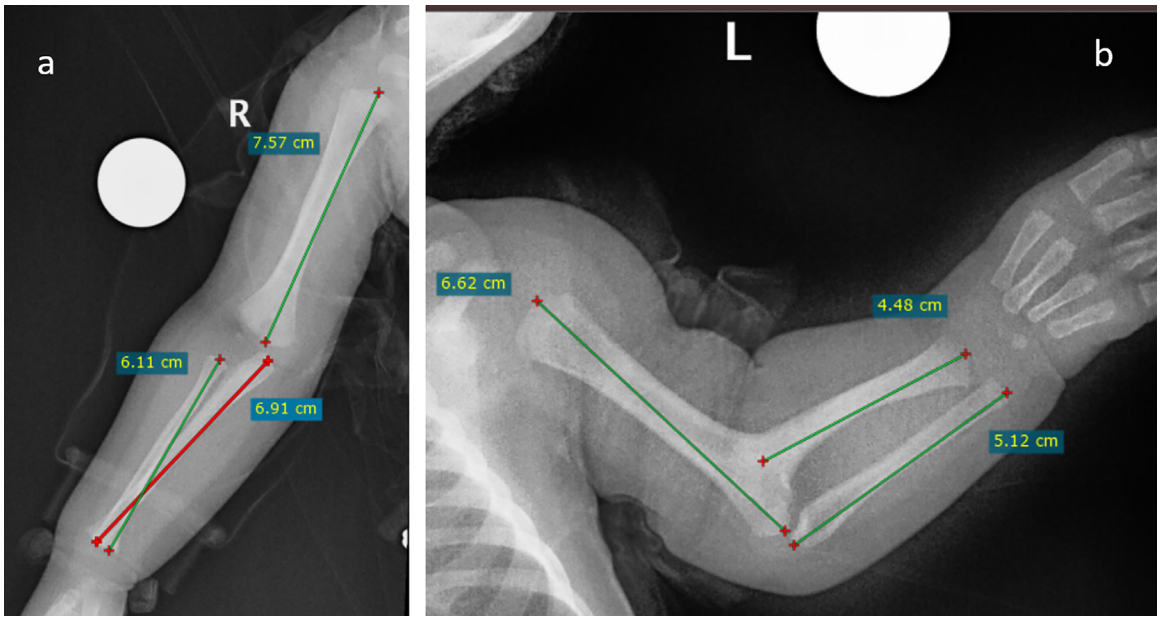

Fig. 2 – Two plain radiographs of the right (image A) and left (image B) upper limbs showing the reduced lengths of the left humerus, radius, and ulnar as compared to the same bones in the right upper limb.

6.9 cm respectively) on the radiographs taken. The distal end of the radius ends 2.0 mm proximal to the distal end of the ulna but both the radius and ulna were not bowed. The ossification center for the left capitate is poorly visualized, unlike the right capitate (Fig. 2).

After a careful review of the patient's information, the orthopedic surgery team decided on conservative treatment as the majority do well according to the literature.

Discussion

CHRS is a rare musculoskeletal condition where there is an abnormal fusion of the bones that make the elbow joint [3]. Even though the exact etiology is unknown, it has been linked to factors such as genetic contributions and cocaine abuse [4]. The infant's mother in our case had no history of cocaine use but admitted to alcohol use and exposure to second-hand cigarette smoke for the first 6 weeks of her pregnancy. CHRS occurs when the separation and retention of the cartilaginous connection between the bones that make the joints of the elbow fails, as it undergoes endochondral ossification [11].

There are 3 known types of synostoses at the elbow, that is, humeroradioulnar, humeroulnar, and humeroradial synostoses, with humeroradial being the commonest [1]. CHRS is further divided into 2 types, class I which is mostly sporadic and associated with ulna hypoplasia, as well as elbow extension, and Class II which is mostly familial and associated with elbow flexion, an isolated upper extremity anomaly, and absence of ulnar ray hypoplasia [5]. However, other reports suggest that familial cases can also present with abnormalities in other organ systems and multiple synostosis syndromes [6,12].

The infant in this case report appears to be a rare sporadic case of class II CHRS presenting with a flexed elbow, shortening of the left radius, ulna and humerus (micromelia), a poorly developed or absent capitate and absence of the left thumb and first left metacarpal. There was no evidence of a syndromic association or the presence of ulnar ray hypoplasia. Early diagnosis and discussion of the findings with the parents are important as some patients have presented to the hospital with radial fractures from attempts by the parents to straighten or mobilize the stiff elbow joint [5]. Fortunately, the infant in our case report had not suffered any fracture.

Treatment for CHRS is either conservative or surgical, the latter being offered to older children which may increase the degrees of freedom at the elbow joint. The majority of patients are reported to do well on conservative treatment, and there is insufficient data to prove surgery is a superior treatment method [5,7].

Conclusion

Sporadic class II congenital humeroradial synostosis is a very rare condition that may present with limitation of elbow joint function. Appropriate timely diagnosis and counseling are required since CHRS is mostly managed conservatively according to literature and may prevent avoidable fractures of the radius from attempts by parents to straightened the flexed fixed elbow, and finally offer adequate time for delayed surgical intervention which is usually ineffective and unhelpful.

Authors' contribution

All the authors contributed substantially to the conception, design, drafting of the manuscript, proofreading, and final approval for publication.

Patient consent

Informed consent was obtained from the patient's parents. Confidentiality and anonymity were ensured.

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