ORIGINAL RESEARCH PAPER

Radio-Diagnosis

RADIOGRAPH BASED DIAGNOSIS OF ARTHROGRYPOSIS MULTIPLEX CONGENITA

KEY WORDS: Arthrogryposis multiplex congenita, musculoskeletal, radiograph, paediatric, diagnosis

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ABSTRACT

Arthrogryposis multiplex congenita (AMC) is a rare disorder, presenting with multiple contractures and limb deformities. Various theories have been proposed for the development of this congenital anomaly. The final diagnosis is made based upon the clinical signs and imaging findings. This case report aims to outline the imaging features that can help radiologists to make the diagnosis of AMC through radiographs and skeletal survey. Since, very few cases have been reported in literature, no specific treatment protocols have been outlined. Therefore, antenatal diagnosis based on characteristic imaging findings is crucial for early termination and parental counselling.

I. INTRODUCTION

Arthrogryposis multiplex congenita (AMC) is a rare congenital, non-progressive musculoskeletal disorder, characterized by multiple contractures, joint deformities, and short for age limbs. Multiple theories have been proposed for the development of flexion and extension deformities, the important ones being under development of a particular group of muscles [1], prolonged in-utero immobilization, or vascular insults [2]. While, confirmatory tests like nerve conduction, electromyography and muscle biopsy may be undertaken to determine an underlying nervous or muscular cause [3], the diagnosis is predominantly made on the basis of clinical signs and skeletal survey radiographs. With very few cases reported in literature, no study defines the radiograph based diagnosis of the condition.

This case report outlines the characteristic radiographic findings of AMC that will help radiologists to diagnose the condition accurately.

II. CASE REPORT

A five-month-old male infant presented with multiple congenital deformities vis flexion deformities in the hand, wrist, and clubfoot. Due to flexion deformity in the hand, the grip was weak. The infant was short for age. Previous antenatal ultrasound scans had not been performed, and no significant antenatal history was present. The patient was a term baby, born through normal vaginal delivery at 38 weeks of gestation.

Multiple radiographs of the axial and appendiceal skeleton were obtained for a detailed skeletal survey. The imaging findings combined together were diagnostic for AMC.

The following imaging findings were seen:

Bilateral feet radiographs: Bilateral congenital talipes equino-varus with short and clubbed metatarsals. There is diffuse soft tissue thickening. (*Figure 1*)

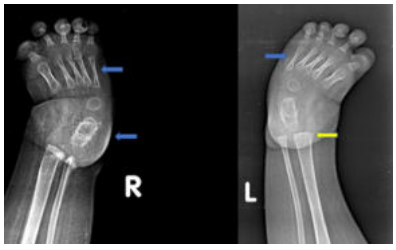

Figure 1

Hand radiograph:

- Short and clubbed metacarpals, and delayed appearance of carpal bones. *(Figure 2)*
- Flexion deformity and Hitchhiker's thumb deformity (Flexion of the first metacarpophalangeal joint with hyperextension of the interphalangeal joint): Seen in hand on the chest radiograph obtained *(Figure 3)*
- Mild ulnar deviation

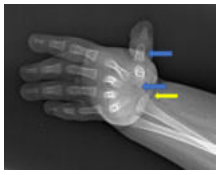

Figure 2

Figure 3

Wrist and ankle: Articular convexity involving the long bones of the wrist and ankle. *(Figures 1,3)*

Spine: No evidence of vertebral body abnormality or myelomeningocele.

- The lower lumbar spine shows anterolisthesis of L5 over S1 vertebral body with spondylolysis at L5 level.
- Mild scoliosis involving the lower dorsal spine with convexity towards the left.

(Figure 4,5)

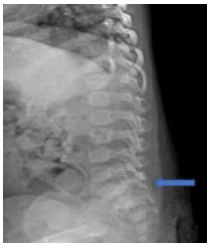

Figure 4

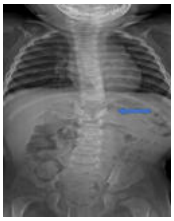

Figure 5

Visualized pelvis and hip joint: Relatively small right femoral epiphysis as compared to the left with disrupted Shenton's line on the right. *(Figure 4)*

Skull Lateral: Absence of posterior clinoid process and embossing of sulci and gyri on the posterior aspect of the skull, suggestive of increased intracranial pressure. *(Figure 6)*

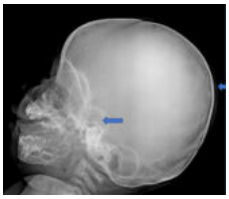

Figure 6

Skull Anteroposterior: Enlarged and protruding pinna with a large-for-face outer ear. *(Figure 7)*

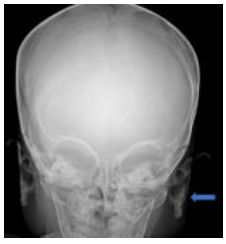

Figure 7

Figure Captions

1. Figure 1: Anteroposterior foot radiographs showing bilateral congenital talipes equino-varus with short and clubbed metatarsals (blue arrows). Articular convexity of long bones (yellow arrow).
2. Figure 2,3: Chest radiograph showing position of hand (3) and anteroposterior radiograph of left hand (2) shows short and clubbed metacarpals with absence of carpals, flexion deformities and contractures at wrist, Hitchhiker's thumb and ulnar deviation at wrist joint (blue arrows). Articular convexity of long bones (yellow arrow).
3. Figure 4: Lateral Spine shows anterolisthesis of L5 over S1 vertebral body with spondylolysis at L5 level.
4. Figure 5: AP radiograph of abdomen, pelvis and hip joint shows relatively small right femoral epiphysis as compared to the left with disrupted Shenton's line on the right. Mild scoliosis involving the thoracic spine is seen.
5. Figure 6: Skull Lateral shows absence of posterior clinoid process and embossing of sulci and gyri on the posterior aspect of the skull, suggestive of increased intracranial pressure.
6. Figure 7: Skull frontal view shows enlarged and protruding pinna with a large-for-face outer ear.

I. DISCUSSION

There are two types of AMC: Amyoplasia and distal arthrogryposis [3]. Clinical features are usually mixed, however, most common is the presence of multiple contractures in various parts of the body. Amyoplasia is the more common variety, and presents with multiple contractures, joint abnormalities as describes below, and severe clubfoot. Patients usually have normal intelligence. Distal arthrogryposis presents with multiple contractures, less involvement of proximal joints, and varied expressivity in association with other syndromes [3].

It can lead to fetal akinesia deformation sequence, because proper fetal growth is dependent on fetal movement [4,5]. There is muscle weakness and hypotonia with weak tendon reflexes. Skin and subcutaneous tissues are flabby and wrinkled. There can be luxation and subluxation of multiple joints [1]. Findings are usually symmetrical and bilateral, and involvement of other organs and systems can be seen.

On imaging:

Antenatal ultrasound:

Reduced fetal movements in utero in second and third trimesters, abnormal fetal position, fixed flexion deformities, micrognathia, altered amniotic fluid volume, limb deformities, cerebral ventriculomegaly, dysmorphic features, and growth retardation [4]. Associated findings like cystic hygroma [6], fetal seizures [7], small chest, thin ribs and multiple diaphyseal fractures [8].

Radiographic findings:

Joint involvement in the form of internal rotation of shoulder, extension / pronation of elbow, volar or ulnar deviation of the wrist, stiff and immobile ankle and wrist joints, bilateral clubfoot, and multiple contractures are noted.

No study outlines a management protocol for the disease yet. However, antenatal diagnosis should be followed by parental counselling regarding the postnatal outcomes.

Teaching point:

In patients presenting for skeletal survey or for bone age estimation, it is important to rule out syndromic, congenital disorders by putting together the skeletal findings.

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