

Case Report:

Rare Case Series of Arthrogryposis Multiplex Congenita

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Abstract:

Arthrogryposis Multiplex Congenita is a term with various etiologies and many complex clinical features like multiple joint contractures of various limb joints.It is associated with malformations,malfunions and neurologic deficiencies.We report the case of a new born, term female child admitted at Rural Medical College,loni(Maharashtra) with positive family history and evident clinical features of arthrogryposis multiplex congenita.Multidisciplinary management was instituted.This is a rare case report

Keywords: Arthrogryposis Multiplex Congenita(AMC), multiple joint contractures, malformations

INTRODUCTION:

Arthrogryposis multiplex congenita is a rare heterogeneous congenital disorder, characterised by non-progressive, multiple,intra-articular contractures that can be recognised at birth, involving more than one area of the body. More over this are more clinical findings than laboratory , associated with different disorders like neuro-cognitive delay and malformations.It occurs due to fetal akinesia which may be because of multiple factors like connective disorder, intrauterine compression, a vascular insult / teratogenicexposure^(2,3)

Symptoms of some forms of arthrogryposis can be found in selected genetic diseases like autosomal recessive,autosomal dominant. Antenatal ultrasound examination can establish the correct diagnosis⁽⁵⁾. 90 % of the cases are associated with birth defects reduction of fetal movements in last month of pregnancy is important indication of these cases. Movement is essential for the normal development of joints and the periarticular tissues⁽⁶⁾. limitation of movements lead to more development of the peri-articular connective tissue

CASE REPORT: 1

A child with multiple deformities with contractures, born to a 26 year old female with G₃P₃L₂D₁ , 2nd degree consanguineous marriage ,was brought to us 6 hours after birth. Her first child died on the 1st day of life with same features as of this child. She has a normal live 2nd child of 4 years of age.Her past medical history was uneventful. Her husband was healthy. No family history of any congenital disorders,with no history of drug use.According to mother decrease movement of baby and ultrasonography there was oligohydroamnios with riding of sutures with cerebral hypoplasia. Neck,chest,spine and abdomen was normal.Bilateral fixed flexed deformity noted at elbow, wrist and interphalangeal joints. Lower limbs were extended with everted foot persistently. Based on these findings diagnosis made and Patient was advised to terminate the pregnancy but the patient continued pregnancy and she delivered at 38 weeks of gestation in a

general poor condition with a low apgar score.Patient was referred to us after 6 hours of life with birth weight of 2.6kg and length of 54 cm and head circumference of 35cm.

On examination after birth, baby showed contractures of the joints typical for AMC i.e affecting elbow,shoulder,wrist,feetand there was knee joint subluxation and dislocation of hip joint.Baby had no sucking reflex and poor reactivity to external stimuli with reduced muscle tone and weak tendon reflexes, X-RAY image there was deformation.Sonography done for both neuroand abdomen butno abnormality was seen.Echocardiography showed moderate sized PDA of size 2.5mm.MRI was not performed due to financial issues. Torch report came to be negative

In this case,more than one type of joints were involved, with specific position and symmetric limb involvement showing the feature of typical AMC.

The child was kept for 10 days and was referred to higher centre for rehabilitation and orthopedic procedures.



Photo 1: Showing contractures of the joints typical for AMC

CASE REPORT: 2

A child with multiple deformities with contractures, born to a 19 year old female with G₁P₁L₁, 2nd degree consanguineous marriage was brought to us on 18 hours after birth. Her past medical history was uneventful. Her husband was healthy. No family history of any congenital disorders, with no history of drug use. In mother ultrasonography there was oligohydroamnios. Bilateral fixed flexed deformity noted.

On examination after birth, baby showed contractures of the joints typical for AMC

i.e affecting elbow, shoulder, wrist, feet and there was knee joint subluxation and dislocation of hip joint. Baby had no sucking reflex and poor reactivity to external stimuli with reduced muscle tone and weak tendon reflexes, X-RAY image there was deformation. Sonography done for both neuroand abdomen but no abnormality was seen. Patient died on third day of life.

In this case, more than one type of joints were involved, with specific position and symmetric limb involvement showing the feature of typical AMC.



Photo 2: Showing contractures of the joints typical for AMC

CASE REPORT: 3

A child with multiple deformities with contractures, born to a 20 year old female with G₁P₁L₁, 3rd degree consanguineous

marriage was brought to us on 48 hours after birth. Her past medical history was uneventful. Her husband was healthy.

Mother was not ANC registered and it was a home delivery.

On examination after birth, baby showed contractures of the joints typical for AMC i.e affecting elbow, shoulder, wrist, feet and there was knee joint subluxation and dislocation of hip joint. Baby had sucking reflex and poor reactivity to external stimuli with reduced muscle tone and weak tendon

reflexes, X-RAY image there was deformation. Abdomen sonography-suggestive of renal dysplasia.

In this case, more than one type of joints were involved, with specific position and symmetric limb involvement showing the feature of typical AMC. On 4th day of life patient was discharged against medical advice.



Photo 3: Showing contractures of the joints typical for AMC

DISCUSSION:

It occurs in 6.2/100000 live births⁽⁷⁾. It is not a specific diagnosis but rather made on the basics of clinical findings. It is associated with more than 300 different

disorders⁽¹⁾. It is seen that anything that inhibits normal joint movement before birth can result in joint contractures.⁽⁸⁾

Arthrogryposis could also be caused by genetic and environmental factors.⁽⁸⁾ The

exact causes of arthrogryposis are unknown yet. Many factors can affect the arthrogryposis they can be intrinsic or extrinsic like in extrinsic factors such as: decreased intrauterine movement, oligohydr amnios , and defects in the fetal blood supply, hyperthermia, limb immobilization and viral infection leads to arthrogryposis⁽⁹⁾. The major cause in humans is fetal akinesia. Another reason can be molecular basis.

It has shown that there are more than 35 specific genetic disorders associated with arthrogryposis these can be chromosomal disorders (for example: trisomy 18).^[10] , mitochondrial or single gene. This is mostly seen in distal arthrogryposis There could also be connective tissue, neurological and muscle development disorders.^[11]

When there is loss of muscle mass with an imbalance of muscle power at joints can also cause this type of abnormalities and myopathies and dystrophy can occur due to dystropin – gycoprotiendysfunction, neurological abnormalities are responsible for more than 80% of the cases of AMC

DIAGNOSIS

The diagnosis can be made prenatally in approximately more than 50% of fetuses presenting arthrogryposis. It could be found during routine ultrasound scanning showing a lack of mobility and abnormal position of

the foetus. We can also use like 4D ultrasound.^[9] , clinically a child can be diagnosed with arthrogryposis with physical examination, confirmed by ultrasound, MRI, or muscle biopsy.

TREATMENT

The treatment of AMC based on patient condition and varies person to person, Early in life, physical therapy to stretch contractures can improve the motion of affected joints and prevent atrophy. Splints can also be used in combination with these stretching exercises. most of the time physical and occupational therapy have proven very beneficial in improving muscle strength and increasing the range of motion of affected joints. Some patients, however requires surgery to achieve better positioning and increase the range of motion in certain joints.^(12,13,14)

PROGNOSIS

The long-term prognosis with AMC depends on the severity of the condition, and person to person with type of involvement and treatment taken. In general, many people affected by AMC have a good prognosis. With physical therapy and other available treatments, substantial improvement in joint function and mobility is normally possible. Most people with AMC are of normal intelligence and are able to lead productive, independent lives as adults.^(15,16)

CONCLUSION:

1. The diagnostic technique of choice is antenatal sonography. Earlier diagnoses influences the influence for termination of pregnancy.
2. Rehabilitation of a child with arthrogyrosis should be early

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