ORIGINAL ARTICLE

Clinical Evaluation of Children with Arthrogryposis Multiplex Congenital in Pakistani Population

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ABSTRACT

Background: Arthrogryposis multiplex congenita (AMC) is a rare syndrome with multiple joint contractures. The number of studies which have been previously done on Pakistani population to investigate patients with arthrogryposis is limited.

Aim: This study presents our experience with evaluation and management of children with AMC at The Children Hospital & the Institute of Child Health, Lahore.

Method: During the period from January 2018 to December 2018, we evaluated 25 children with AMC. The mean age at the time of evaluation was 20.56 months (range, 5 days to 9 years). All of the included patients were evaluated in terms of their clinical state according to selected subjective and objective criteria.

Result: Our results indicate that shoulders were internally rotated (20%), the elbows were extended (36%), and the wrists (76%) and digits (56%) were flexed. Thumb was adducted (36%), hands were clawed (52%) and palmar crease was absent (28%). Hips were found to be dislocated (16%) and adducted (16%). Knees having extension contracture (32%); feet were in equinovarus (72%). Calf muscles were atrophic (56%).

Conclusion: Our experience shows that the clinical findings are highly variable between families and also within families with AMC. Joint manipulation and casting during the first few months of life may produce considerable improvement. Orthotics may help. Surgery may be needed later, but mobility is rarely enhanced. Muscle transfers may improve function. Many children do remarkably well; two thirds are ambulatory after treatment. A multidisciplinary team evaluation of the child with AMC is recommended for specific diagnosis and planning of treatment.

Keywords: Arthrogryposis multiplex congenita (AMC), clinical evaluation.

INTRODUCTION

The term arthrogryposis is a misnomer; its simple meaning is "stiff joints". We use this term when more than one joint is involved. It's actually a syndrome including different diseases that have common presentation of joint stiffness, visceral and neurological involvement. Very limited work is available in literature regarding its clinical presentation in Pakistan. Its incidence varies from 1/3000 to 1/5100 live births (1). Exact etiology is not known, but it is thought secondary to fetal joint development, due to multiple abnormalities of neurological, muscular, vascular, connective tissue disorders or some environmental factors (2). Multiple types of arthrogryposis are described in literature including, Amyoplasia, distal arthrogryposis and syndromic arthrogryposis. Although association of certain genes with few types of arthrogryposis is established with advances in research in molecular genetics field, but still a lot of thing are still missing in this regard. Gene mutation and their mode of transmission is identified in few types (3,4). Due to this diverse nature of disease and not knowing its exact etiology we could not have a standard treatment protocol for each patient up till now. Each patient varies due to its different presentation and involvement of different organs. So we mainly focus on improvement of functional status of each limb and patient mobility status as a whole. Our goal of this study was to show the typical presentation of arthrogryposis kids in Pakistan, frequency of associations with different organs in same patient.

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MATERIAL AND METHOD

From January 2018 to December 2018, 25 children were identified for arthrogryposis in the Outdoor department of Paediatric Orthopaedic, Children's Hospital and Institute of Child Health Lahore. Our inclusion criteria included all children presenting with joint stiffness present at birth and involving at least two or more joints. Children who have isolated club foot deformity, congenital vertical talus and joint stiffness associated with condition such as cerebral palsy and polio were excluded from the study. After informed consent from the parents, demographic profile (age and sex) was noted. All relevant data relating to family history, pre and perinatal history was taken from mother. Our clinical examination included a Performa noting the general features of a child, his posture, presence of contracture to the specific joint of the limb and spine involvement. Involvement of all four extremities, short limb, clenched fist, camptodactyly, craniofacial abnormalities, ophthalmoplegia, claw hand, whistling face, wing like web of knee and elbow and web neck was noted as general features. We also examined the posture of child as having either extended elbows, flexed wrist, ulnar deviation of hand, extended knees, equino-cavo varus feet, congenital vertical talus, atrophic arm and legs and presence or absent of joint crease. Then, we noted the presence or absence of contracture to the specific joint of the upper and lower limb. Spine examination is also performed to note the presence of any deformity like scoliosis. Those who had bilateral involvement were counted as having two separate features. All data was recorded and maintained. Quantitative variables like age (in months), mean±SD. Qualitative

variables like gender, birth history, general features, posture and specific joint deformity was presented in the form of frequency and percentages.

RESULTS

The mean age of the children included in our study was 20.56 ± 6.78 months and there were 16 (64%) males and 9 (36%) females. 12 (48%) were born via C-sections while 12 (52%) were born via SVD. 5 children had brother or sister as having the same symptomatology and cousin marriage was noted in 8 children. Pregnancy related complications like pre-eclampsia, oligo-amnios, bleeding was noted down from mothers and we observed 7 children born with any one of these associated complications. There was no twin pregnancy. Presentation was predominantly cephalic (70%) as evident on ultrasonography. The delivery was term in 20 children and premature in 5 and the weight was around 2.2 pounds in 14 children. Clinical examination of the child revealed involvement of all four extremities in 17(68%), short limb in 2(8%), clenched fist in 9 (36%), compatodactyly in 14(56%), craniofacial abnormalities in 3(12%), ophthalmoplegia in 3(12%), claw hand in 13(52%), whistling face in 3(12%), wing like web of knee in 5(20%) and elbow in 2(8%) and web neck in 2(12%). Posture with extended elbow was seen in 9(36%) children, flexed wrist along with ulnar deviation of hand in 19(76%). The most common lower limb posture was extended knees (32%) in the lower limb. Atrophy of leg and forearm was seen respectively in 8(32%) and 12(48%). Absent palmar crease was seen in 7(28%). Joint examination revealed flexion contracture of the elbow, wrist, thumb and fingers as most common in the upper limb. In lower limb, hip involvement as DDH (16%) and adductor contracture (16%) was seen. Knee as hypertension deformity (44%) and foot as CTEV (64%), CVT (52%) were the notable deformities. In spine, we found scoliosis in 4(16%) and lordosis 3(12%) children.

Table: Features of Arthrogryposis multiplex congenita (n=25).

	n	%age
Age	20.56 months (Average)	
Gender		
Male	16	64%
Female	9	36%
Clinical History		
C- Section	12	48%
General Features		
All four extremities involved	17	68%
Short limbs	2	8%
Clenched fist	9	36%
Compatodactyly	14	56%
Craniofacial abnormality	3	12%
Ophthalmoplegia	3	12%
Claw hand	13	52%
Whistling face	3	12%
Wing-Like Web of Knee	5	20%
Wing-Like Web of Elbow	2	8%
Web Neck	3	12%
Posture		
Extended Elbow	9	36%
Flexed wrist	19	76%
Ulnar Deviation of Hand	19	76%

Knee Extended	8	32%
Equino-cavo-varus foot	18	72%
Atrophic arms	8	32%
Atrophic legs	12	48%
Absent joint crease	13	52%
CVT	13	52%
Shoulder		
Internal Rotation Contracture	5	20%
Elbow		
Flexion Contracture	1	4%
Extension Contracture	2	8%
Wrist		
Extension Contracture	2	8%
Flexion Contracture	11	44%
Absent palmar crease	7	28%
Thumb		
Opponence Contracture	9	36%
Fingers		
Flexion Contracture	14	56%
Hip		
Dislocation (DDH)	4	16%
Adduction Contracture	4	16%
Knee		
Hyperextension	11	44%
Flexion Contracture	5	20%
Extension Contracture	5	20%
Feet		
CTEV	16	64%
Calf Atrophy	14	56%
Absent Flexion Crease	5	20%
Pes Planus (Bilateral)	2	8%
Hallux Valgus	2	8%
Spine		
Scoliosis	4	16%
Lordosis	3	12%

DISCUSSION

Arthrogryposis is not a diagnosis rather it is a group of clinical findings. According to some of studies it is present in more than 100 syndromes. Regarding epidemiology its prevalence has very diversity from 1/3000 to 1/56000 live births. This diversity may be due to lack of a clear-cut diagnostic criterion that is unfortunately not available up till now. Arthrogryposis is evident at birth and mostly it involves all four limbs, but it can present with involvement of a single limb, although it is rare presentation. Most common deformity is clubfoot. Almost 90% cases present with this problem. This deformity may be due under-developed or absent muscles or due to fibrosed tissues of joint, exact cause not known. This deformity is resistant to routine Ponsity casting and usually need more number of casts than needed in idiopathic clubfoot. Other foot deformities include mainly cavus, equinus, calcaneo-valgus or any combination. Hip involvement is also very common. Scissoring or hip adduction contracture can leads to hip subluxation and ultimately hip dislocation. Passive hip abduction is achievable in majority of cases at the time of birth. If hip is located then flexion-abduction-external rotation contracture is common, but if hip is dislocated then it is in position of fixed adduction. Shoulders also present usually in adduction and internal rotation contracture, with limited range of motion. Elbow presented with flexion or

extension contracture and forearm in pronated position. Wrist in flexion attitude along with flexion contracture of all digits at PIPs and DIPs joints. Thumb in palm deformity is common. Spine deformities are common and incidence varies between 2.5% to 65.9%(10). Anomalies other than bone and joints include; characteristic facial features, rounded expressionless face¹¹, hemangiomas spot on face, genito-urinary, gastrointestinal, cardio-vascular or maxillary abnormality (micrognathia)¹². Incidence of these extra-osseous features are comparatively rare.

There are many types of arthrogryposis but congenital amyoplasia is most frequent type (one third of cases)^{8,13}. It is characterized by symmetrical muscle hypoplasia and associated distal limb deformities. Muscles are usually replaced by fibers and adipose tissue. Distal arthrogryposis is a particular type of arthrogryposis affecting hand and feet¹⁴.

Exact etiology of arthrogryposis is not known, but there are few findings. Absence of fetal movements, fetal akinesia appears to be basic cause. Earlier this happens in intrauterine life, more severe the presentation at birth³. It is observed that joint development remains normal during embryogenesis, but due to increase thickening of joint capsule and associated defective muscles, joint stiffness is exaggerated^{3,15}. Majority of cases of Arthrogryposis have associated neurological abnormalities (90%) and most common is decrease in cells in the anterior horn of spinal cord¹⁶. Other neurological association includes peripheral neuropathy, neuro-muscular junction transmission failure^{3,17}. Some studies correlate arthrogryposis with maternal and metabolic abnormalities^{3,18}. Diagnosis of classification of arthrogryposis is important for better treatment and prevention of recurrence^{3,4}. Pre-natal screening can detect joint stiffness, presence of deformities and decrease fetal movements, oligohydramnion, polyhydramnion by Ultra sonography, even at 16th to 18th week of gestation^{6,19}. Nuchal translucency detection is possible in third trimester²⁰. Prenatal diagnosis can help to manage delivery at better place so to reduce expected birth complications at time of delivery.

Main things that help to maintain and increase joint movement are rehabilitation, orthosis and surgery⁴. Rehabilitation gives good results if started at birth because joints are very flexible at that time. But it must be gentle to prevent fracture and plastic deformation of bones should started at birth is very important to increase and maintain joint movements. Casting can help to correct deformities of feet and hands. In upper limb we mainly concerned regarding functional capability of patient like eating, cleaning, washing, holding crutches etc. so role occupational therapy is very important. Role of surgery for upper limb is very rare, as many patients become able to perform their daily activity of living by alternate movements of upper limb^{21,22}.

Surgery is more common for lower limb deformities. Hip dislocation or even subluxation is indication of surgery, open reduction or soft tissue contracture releases for correction of hip subluxation, stiffness, avascular necrosis of head of femur^{9,23}. For genu recurvatum casting gives better results if started in early life but surgical release of soft tissue or even osteotomy can be an option for resistant cases. If hip and knee both effected then treatment started

addressing knee first²⁴. To address knee flexion contracture, serial knee extension casting gives good results but if contracture is more severe then surgical releases of hamstrings and posterior capsulotomy of knee joint or even femoral shortening and gradual correction by using illizrov, can be an option^{25,26}.

For clubfoot deformity in arthrogryposis, Ponsy casting should be first choice. These feet are more resistant as compared to idiopathic clubfoot²⁷. Even though it involves more number of casts, relapses and increase number of other complications; but surgery should be last option. Surgical releases should be thorough so to prevent recurrences as it would lead to stiff foot and more chances of vascular injury in subsequent surgery²⁸. Severe deformed foot can be corrected by illizrov fixator or by talectomy procedure²⁹. Surgical release of Cavus foot also have good results³⁰. Arthrogryposis patients also need special precautions regarding anesthesia. Direct laryngoscopy and Intubation can be difficult due to small jaw or narrow airways³¹. These patients are sometime more sensitive to respiratory depression effect of analgesic drugs. Breathing difficulties can be due to myopathy, pulmonary hypoplasia or spinal deformity¹¹. PODCI score is one of good tool to assess the function, happiness and pain in children with limitations in joint mobility⁵. The prognosis depends mainly on etiology diagnosis and proper rehabilitation²². Congenital stiffness in distal arthrogryposis is easier to treat than in amyoplasia that usually require surgeries.

CONCLUSION

As the causes of arthrogryposis are not curable, the goal of support is to improve the functions of bulation and prehension and acquire the maximum of self-nomie to facilitate social integration and ensure education normal. The role of the family for the integration of the child in society is very important. Psychological care logic of the parents is part of the treatment because they must be able to withstand a long treatment starting from birth and continues throughout growth.

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