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Original Research Article

A study on joint involvements of the children suffering from arthrogryposis multiplex congenita attended at a tertiary care centre of eastern India

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ABSTRACT

Background: Arthrogryposis multiplex congenita (AMC) is a disorder which present at birth and characterized by multiple joint contractures and defective muscles, with normal sensations. This condition was first described by Otto in 1841. In 1923 Stern first used the term AMC in 1932. The objective of this review is to summarize the demography related to arthrogryposis, especially its age of presentation, gender predominance, joints involved, whether both upper limb and lower limb or only lower limb affected.
Methods: Total 30 patients in the age group new born to 18 years included in the study. It is a combination of retrospective and prospective observational study. The following parameters are analysed in the study, Age at presentation, gender, type and severity of joints affected.
Results: In our study, 8 (26.7%) patients were <12 months old, 13 (43.3%) patients were 12-60 months old, 8 (26.7%) patients were 61-120 months old and 1 (3.3%) patient was >120 months old. 9 (30.0%) patients were female and 21 (70.0%) patients were male. 27 patients have combined upper and lower limb involvement and 3 patients has only lower limb involvement. In our study, 2 (6.7%) patients had progressive upper limb and 28 (93.3%) patients had regressive upper limb.
Conclusions: Most common joints which are affected in AMC are shoulder, elbow, wrist, MCP, IP, hip, knee, ankle and the most common position of these joints are internal rotation of shoulder, extended elbow joint, flexed wrist joint, extension of MCP joints, extension of IP joints, flexed hip joint, flexed knee joint and presence of club foot and hip dysplasia (DDH).

Keywords: Arthrogryposis multiplex congenita, Joints, Contracture, Club foot, DDH

INTRODUCTION

Arthrogryposis multiplex congenita (AMC) is a disorder which present at birth and characterised by multiple joint contractures and defective muscles, with normal sensations.[1] This condition was first described by Otto in 1841. In 1923, Stern first used the term AMC.[2] In 1932, the specific name of amyoplasia was given by Sheldon. The most common feature is the lack of foetal movements.[3] This can result from a large number of disorders, including neuropathies, abnormalities of the muscles, connective tissue disorders, conditions that limit the internal space of the uterus or defects of the uterine environment.[4,5] Because there are 150 different disorders or syndromes that have joint contractures as part of their manifestations, AMC is a diagnosis of exclusion6, and making a precise diagnosis is very important for the evaluation of results. Arthrogryposis (arthrogryposis multiplex congenita - AMC) is not a separate disease entity, but is rather a descriptive diagnosis with varying

etiologies.[7,8] Their common feature is the presence of congenital, usually progressive or non-progressive joint contractures involving at least two different body areas.[9] This class of diseases includes the so-called classic arthrogryposis - amyoplasia, with its unique clinical features such as symmetrical, severe contractures, usually involving both the upper and lower limbs .Most contractures in arthrogryposis require treatment; this potentially involves multiple surgical corrections of the knees and hips, correction of spinal deformities, elbow and wrist contractures, foot deformities, and of dislocations commonly accompanying the contractures; of these, the hip and knee are the most commonly affected joints.[10,11] The objective of this review is to summarize the demography related to arthrogryposis, especially its age of presentation, gender predominance, joints involved, whether both upper limb and lower limb or only lower limb affected. The above-mentioned joint abnormalities may involve all limbs (usually) in the lower or the upper extremities. Arthrogryposis is usually symmetrical, but, less frequently, various joints may be involved to a different extent.[12,13]

METHODS

It is a combination of retrospective and prospective observational study. Study was conducted in KPC medical college, Kokata, West Bengal from March 2017 to December 2020. Total 30 patients in the age group new born to 18 years included in the study. The following parameters are analysed in the study, age at presentation, gender, type and severity of joints affected. Selection criteria of the patients are the followings, new born children and children aged up to 18 years, children with congenital contracture of 2 or more joints and the parents or guardian of the children who will give the written consent were included in the study. Ethical approval is taken from the institution ethical committee.

Sample size has been calculated with help of Epi Info (TM) 7.2.2.2. EPI INFO which is a trademark of the Centres for Disease Control and Prevention (CDC).

As per the available information from the Department of Medical Records of KPC Medical College 81.42% the patients suffering from AMC were with age <12 years.

Thus, for this study p=0.8142. The number of patients required for this study will 30.49~30 with power 83%. The formula used for sample size calculation is as follows.

n= Required sample size

p= 0.8142 as per the available information in the Department of Medical Records

q = 1 - p

L = Loss % (Loss of information) = 17.3~17%

Calculation

Here p=0.8142, q=1-p=0.1858, Loss % = 17.3%

4pq = 4 X 0.8142 X 0.1858 = 0.6051

L2 = (0.8142 X 0.173)2 = 0.0198

So, n= 0.6051 / 0.0198=30.49 ~ 30

The following parameters will be included in the proposed study: age at presentation, gender, all the joints of upper limb and lower limb affected with their positions, type and severity of the most frequent contractures at birth, whether partly treated or untreated during their 1st visit in this institute, and natural course of disease of upper limb and lower limb.

Statistical analysis

For statistical analysis data were entered into a Microsoft excel spreadsheet and then analyzed by SPSS (version 27.0; SPSS Inc., Chicago, IL, USA) and GraphPad Prism version 5. Data had been summarized as mean and standard deviation for numerical variables and count and percentages for categorical variables. Two-sample t-tests for a difference in mean involved independent samples or unpaired samples. Paired t-tests were a form of blocking and had greater power than unpaired tests. One-way analysis of variance (one-way ANOVA) was a technique used to compare means of three or more samples for numerical data (using the F distribution). A chi-squared test ($\chi 2$ test) was any statistical hypothesis test wherein the sampling distribution of the test statistic is a chi-squared distribution when the null hypothesis is true. Without other qualification, 'chi-squared test' often is used as short for Pearson's chi-squared test. Unpaired proportions were compared by Chi-square test or Fischer's exact test, as appropriate.

Explicit expressions that can be used to carry out various t-tests are given below. In each case, the formula for a test statistic that either exactly follows or closely approximates a t-distribution under the null hypothesis is given. Also, the appropriate degrees of freedom are given in each case. Each of these statistics can be used to carry out either a one-tailed test or a two-tailed test.

Once a t value is determined, a p-value can be found using a table of values from Student's t-distribution. If the calculated p-value is below the threshold chosen for statistical significance (usually the 0.10, the 0.05, or 0.01 level), then the null hypothesis is rejected in favour of the alternative hypothesis. P value ≤ 0.05 was considered for statistically significant.

RESULTS

In our study, 8 (26.7%) patients were <12 months old, 13 (43.3%) patients were 12-60 months old, 8 (26.7%)

patients were 61-120 months old and 1 (3.3%) patient was >120 months old (Table 1). In our study, 9 (30.0%) patients were female and 21 (70.0%) patients were male (Table 2), 27 (90.0%) patients had combined AMC and 3 (10.0%) patients had distal AMC.

Table 1: Distribution of AMC according to age in months.

Age in months	Frequency	Percentage
<12	8	26.7
12-60	13	43.3
61-120	8	26.7
>120	1	3.3
Total	30	100

It is found that hip, knee, shoulder and elbow joints are most commonly affected joint, out of 30 patients 23 patients has shoulder internally rotated, and 19 patients has bilateral shoulder involvement. 23 patients elbow affected out of which 19 has in extended and 4 has flexed position. 2 patients had extended wrist joint, 20 patients had Flexed Wrist.19 patients has interphalangeal joint involvement.

Table 2: Distribution of AMC according to sex.

Sex	Frequency	Percentage
Female	9	30
Male	21	70
Total	30	100

All thirty patients have hip involvement out of which 6 has developmental dysplasia of hip (DDH). Most of patients has fixed extension deformities of knee of which 5 has congenital dislocation of knee (CDK). Twenty-eight patients have congenital talipes eqinovarus deformity of foot. Overall joint involvement is evaluated in Table 3. In our study, 10 (33.3%) patients had progressive lower limb and 20 (66.7%) patients had static lower limb deformities. In case of upper limb 2 (6.7%) patients had progressive upper limb and 28 (93.3%) patients had regressive deformities.

Table 3: Distribution of joint affected, laterality and deformities in AMC.

Joint involved	No. of patient	Bilateral involvement	Position of joint
Shoulder	23	19	Internally rotated-23
Elbow	23	20	Internally rotated-19
			Externally rotated-4
Wrist	22	20	Flexed-20
			Extended-2
Metacarpophalangeal	27	27	Extended-26
			Flexed-1
Interphalangeal	27	27	Extended-26
			Flexed-1
Hip	30	28	Flexed/externally rotated-28
			Extended-2
Knee	26	24	Flexed-20
			Extended-6
Ankle and foot (CTEV)	28	27	Foot equinous, adducted and varus
DDH	11	2	Hip shortened and externally rotated
CDK	25	24	Knee extended

DISCUSSION

In developing or under developed countries a child born with so many deformities are considered a curse of god and often remains neglected. Even when the parents are keen to have proper treatment and training for their challenged children, only a few will have access to the limited number of centres of excellence that may exist in these countries. The purpose of the current study was to describe the natural history of disease. Since there are no established diagnostic criteria for amyoplasia, many studies may include children with amyoplasia as well as other MCC conditions, which would change the results. An accurate diagnosis is essential for understanding the natural history and prognosis for these children.

In our study, 8 (26.7%) patients were <12 months old, 13 (43.3%) patients were 12-60 months old, 8 (26.7%) patients were 61-120 months old and 1 (3.3%) patient was >120 months old. In above table showed that the mean age in months (mean ±SD) of patients were 42.9667±40.9023 which was higher than the finding in the study of Oncel et al with mean age at the first evaluation was 22 months (range: 9 days-180 months).[14] It may be due lack of knowledge of the parents regarding where to go for treatment in such a situation. In our study, 9 (30.0%) patients were female and 21 (70.0%)

patients were male. It was found that male was higher in this study whereas Hall et al in their study found that the sex ratio was found to be 59% female and 41% males.[15] In our study, 27 (90.0%) patients had combined AMC and 3 (10.0%) patients had AMC affecting the lower limb only whereas Hall et al in their study found that 63% of patient had combined AMC. B) 24% with mainly upper limb involvement. C) 13% with mainly lower limb involvement.[16] In our study 73.3% patients had IR shoulder joint position and 26.7% patients had neutral shoulder joint position and in severe difficulty group, 100.0% patient had IR joint position where as in the study of Hall et al 100% patients had internal rotation of the shoulder.[16]

In our study 19 (63.3%) patients had extended elbow, 4 (13.3%) patients had flexed elbow and 7 (23.3%) patients had neutral elbow and in severe difficulty group, 100.0% patient had extended elbow which is similar to the finding in the study of Hall et al where 65% had rigid extension of the elbow and rest had flexion contracture at elbow joint.[16]

Figure 1: 11 months old male child with flexion contracture of hip and knee joints with CTEV.

Figure 2: 80 months old male child with flexion contracture of elbow joint.

Figure 3: 5 months old male patient presented with flexion contracture of elbow and wrist of left side and on the right side with extension at elbow joint and flexion at wrist joint.

Figure 4: 24 months old male child with B/L shoulder internally rotated.

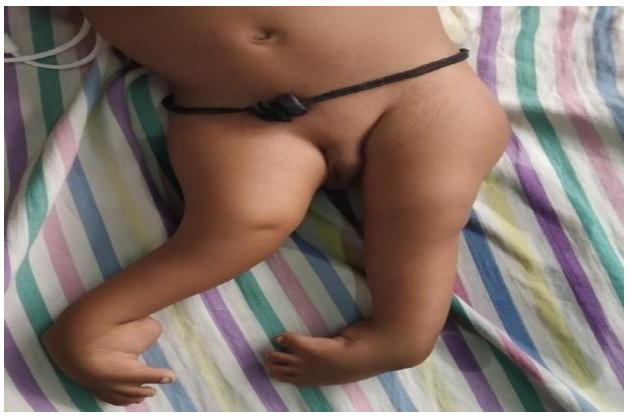

Figure 5: 14 months old female child with B/L flexion deformity at hip and knee joints with B/L CTEV.

In our study 2 (7%) patients had extended wrist joint, 22 (73.0%) patients had flexed wrist joint and 6 (20%) patients had neutral wrist joint and in severe difficulty

group, 100.0% patient had flexed wrist joint which is similar to the finding in the study of Hall et al where 88% had flexion contracture at the wrist joint. Limitation of our study is the number of the patients is less. Further study needed with a greater number of patients to analyse the joint involvement accurately in AMC.

CONCLUSION

Most common joints which are affected in AMC are shoulder, elbow, wrist, MCP, IP, hip, knee, ankle and the most common position of these joints are IR of shoulder, extended elbow joint, flexed wrist joint, extension of MCP joints, extension of IP joints, flexed hip joint, flexed knee joint and presence of club foot. This characteristics involvement of joint and presence of CTEV, DDH. CDK can raise suspicion of AMC in mind of clinician both paediatrician and orthopaedic surgeon. And if we can diagnose and intervene AMC early in childhood the functional outcome in both physical and educational areas will be excellent.

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