

# Walking ability in patients with arthrogryposis multiplex congenita

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## ABSTRACT

**Background:** Arthrogryposis multiplex congenita (AMC) is a multiple joint condition which affects both lower and upper extremities and thus affects ambulation. Multiple surgeries are needed to correct limb deformity in order to promote walking. The objective of this study is to identify the most critical residual deformity that diminishes the ambulatory status.

**Materials and Methods:** 51 patients were included in this study, 14 patients were nonambulatory. The mean age at first surgery was 4.1 years (range 2-16 years). The mean length of followup was 44.0 months (range 22-168 months). Type of procedures and number of operations, residual deformity and walking ability were recorded. Residual deformity including hip flexion contracture more than 30°, knee flexion contracture more than 30°, scoliosis, hip dysplasia or dislocation, knee extension contracture or recurvatum, active motion of hips and knees and upper limb involvement were evaluated. Statistical analysis was done to evaluate factors that were statistically significant to affect walking ability in AMC patients.

**Results:** At the latest followup, 31 patients were community ambulators, 3 patients were household ambulators, 3 patients were nonfunctional ambulatory, and 14 patients were nonambulatory. There were an average of 4.3 surgeries per patient. Statistical analysis of all factors was done and the results were significant with a  $P < 0.037$  in knee flexion contracture  $>30$  degrees with odds ratio of 4.58. Hip flexion contracture  $>30^\circ$  was a trend toward significant with a  $P$  value of 0.058 and odds ratio of 4.53. Multivariate analysis showed that knee flexion contracture was significant with 4.58 (95% CI 1.01-20.6).

**Conclusion:** AMC is a rare disease that causes disability, requiring multiple surgeries to correct deformities. Our study showed that residual knee flexion contracture was associated with nonambulatory status of patients with AMC.

**Key words:** Arthrogryposis multiplex congenita, amyoplasia, knee flexion contracture, walking prognosis

**MeSH terms:** Arthrogryposis, amyoplasia congenita, contracture, knee

## INTRODUCTION

Arthrogryposis multiplex congenita (AMC) is a rare, nonprogressive congenital disorder which is characterized by multiple joint contractures with normal sensation.<sup>1-5</sup> Diagnosis of AMC is usually based on physical examination with characteristic deformities of upper and lower extremities, limited range of motion, clubfoot or hip dislocation. Patients are usually painless

and have normal intelligence. There are 150 different disorders for syndromes that have joint contractures as a part of their manifestations; AMC is diagnosed by exclusion, and precise diagnosis is very important for the evaluation of results.<sup>6</sup> The term “amyoplasia” refers to the most common arthrogryptic syndrome, which includes multiple congenital contractures, typical and symmetrical positioning of the limbs and the replacement of muscles by fibrous and fatty tissues. It is sporadic, with an unknown hereditary pattern.<sup>7</sup> The goal of treatment is to obtain the maximum possible function for each child by an early multidisciplinary approach.<sup>7,8</sup> At present the goal of treatment is to improve ambulatory status of patients and patient self-care. AMC patients have multiple problems which can be improved by surgical intervention but the most helpful procedures are still undefined. This study aims to find residual deformity that affects the ambulatory status of the patient. Residual deformities that are risk factors in this study include hip flexion contracture more than 30°, knee flexion contracture more than 30°, knee extension deformity or recurvatum, severe scoliosis, upper extremity contracture or weakness and development dysplasia of the hip (DDH) or hip dislocation. We feel that our results will

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guide physicians in decision making regarding treatment to promote ambulation in AMC patients.

## MATERIALS AND METHODS

This study was designed as a retrospective study of AMC patients at our institute from January 1995 to December 2010. After approval from the Ethics committee, chart review was conducted in accordance with the guidelines provided from our Institutional Review Board. We reviewed all the medical records and included only those children with AMC who had a mean followup of 44 months (range 22-168 months). The inclusion criteria were AMC patients who underwent surgical procedure or treatment for deformities or dislocations of the joints in the upper or lower limbs and patients or parents who agreed to participate. The exclusion criteria were unclear diagnosis and loss to followup. The study recruited 53 patients; two of them were excluded because of incomplete medical data.

Fifty-one patients were enrolled. We recorded the children's potential to ambulate at the first visit and at each of the subsequent visits until the last followup. All clinical assessment was recorded and included age, sex, associated disease, and surgical procedure carried out. Ambulatory status and residual deformities which include deformities in both upper and lower extremities, scoliosis, degrees of contracture at the hips and knees, active motion of hips and knees and the presence of DDH or hip dislocation were recorded from the clinical and physiotherapy notes. The primary outcome was residual deformity which affected the ambulatory status of the patient.

Statistical analysis using SPSS for Windows and  $P < 0.05$  were considered significant. Multivariate analysis was used to analyze for 95% confidence interval.

## RESULTS

There were 28 females and 23 males. Thirty three patients were in walking age and twenty patients were ambulatory at the start of treatment. Walking age in this study was patient age more than 18 months. All the patients who were ambulatory at the start of treatment remained ambulatory at final followup. Out of thirty one patients who were nonambulatory at the start of treatment, seventeen patients could ambulate at the final followup, fourteen patients were still nonambulatory. Of the thirty one patients who were nonambulatory at the start of treatment, twenty one patients had hips and knee flexion contracture of more than 30° and after proximal femoral extension osteotomy and distal femoral extension osteotomy, seventeen patients who were corrected to less than 30° of hips and knee flexion contractures could walk. For the residual deformities, seven patients had hip

flexion contracture more than 30° and nine patients had knee flexion contracture more than 30°. Forty seven patients were able to do active range of motion of hips and knees. Thirty six patients had upper extremity deformity or weakness. Three patients had scoliosis. Hip dislocation or DDH were seen in 12 patients [Table 1]. All surgical procedures in this

**Table 1: Results**

Variables (N)	Walking	Non walking	Crude odds ratio	P value
<b>Hip FC</b>				
>30 degrees (7)	3	4	4.53	0.058
<30 degrees (44)	34	10	(0.87-23.72)	
<b>Knee FC</b>				
>30 degrees (9)	4	5	4.58	0.037
<30 degrees (42)	33	9	(1.02-20.69)	
<b>Scoliosis</b>				
Present (3)	2	1	1.35	0.999
Absent (48)	35	13	(0.11-16.13)	
<b>Active ROM Hip and knee</b>				
Absent (4)	2	2	0.34	0.300
Present (47)	35	12	(0.04-2.71)	
<b>Knee recurvatum</b>				
Present (6)	3	3	3.10	0.327
Absent (45)	34	11	(0.54-17.59)	
<b>Upper limb weakness/ contracture</b>				
Present (36)	28	8	0.43	0.301
Absent (15)	9	6	(0.12-1.57)	
<b>Dysplasia or Hip dislocation</b>				
Present (12)	6	3	1.41	0.692
Absent (39)	31	11	(0.30-6.62)	

FC=Flexion contracture, ROM=Range of motion

**Table 2: Location and type of operations for the upper limbs**

Site	Type of operation	Number
Elbow	Distal humeral osteotomy	2
	Tricepsplasty and soft tissue release	9
Wrist and Hand	Proximal row carpectomy	17
	Tendon transfer	20
	Hand reconstructions	6
Total		54

**Table 3: Type of operations for the lower limbs**

Site	Type of operation	Number
Hip	Open reduction of the hips and Pelvic osteotomy	11
	Proximal femoral osteotomy	11
	Soft-tissue and tendon release	9
Knee	Distal femoral osteotomy	6
	Quadricoplasty and patellar realignment	13
Foot	Soft tissue release for clubfoot	29
	TAL	37
	Talectomy	39
	Midfoot osteotomy	5
	Triple arthrodesis	5
Total		165

TAL=Tendo achilles lengthening

study were managed by two surgeons. There were 54 upper extremities procedures and 165 lower extremities procedures [Tables 2 and 3]. The average number of operations were 4.3 procedures per child. Bilateral limb surgeries were considered as two procedures, even if they were done during the same anesthesia [Tables 2 and 3]. The final functional outcome was assessed using Hoffer's classification of walking status [Table 4].<sup>9</sup> Thirty one patients were community ambulatory who were able to walk with aids in the community and did not need a wheel chair, three patients were household ambulatory, able to walk with aids in the house and use a wheel chair in the community whereas three patients were nonfunctional ambulators using a wheel chair and capable of transfer while 14 patients were nonambulators and were not capable of transfer [Table 4].

At the last followup, 37 patients were ambulatory and 14 patients were nonwalkers. There was statistical significance with *P* value 0.037 for knee flexion contracture more than 30° with odds of 4.58 (95% CI 1.01-20.6). Hip flexion contracture more than 30° was a trend toward significant with a *P* value 0.058 and odds ratio of 4.53. Other residual deformities were no statistical significant [Table 1].

## DISCUSSION

The management of the musculoskeletal problems associated with arthrogyriposis is challenging. These children are best managed by multidisciplinary teams that include an experienced geneticist, an orthopedic surgeon, a physical therapist, and a pediatric physical therapist. It should be recognized that high percentage of these children might be able to achieve some measure of functional ambulatory potential, but many will lose ambulation as they get older. The need for surgery is high in these children, and often, several procedures will be necessary. A careful discussion of realistic goals with the parents is very important, as is the ongoing focus on skills necessary for promoting independence in adulthood. Hoffer *et al.*<sup>9</sup> reported functional ambulation in 22 out of 36 severely affected adolescents and young adults with arthrogyriposis. Many series have reported varying degrees of functional ambulation in 50 to 78% of patients.<sup>10,11</sup> Sells *et al.*<sup>11</sup> found that 85% of children with amyoplasia were ambulatory by the age of 5. These children had an average of 5.7 orthopedic procedures along with multiple casting, splinting of limbs and regular physical and occupational therapy.

**Table 4: Characteristics of walking at latest followup**

Hoffer's classification	Number
Community ambulatory	31
Household ambulatory	3
Nonfunctional ambulatory	3
Nonambulatory	14

Ultimately they were able to participate in activities of daily living and attend school. Based on our findings we strongly believe that children with arthrogyriposis may have good potential for ambulation if their knee flexion contractures have been adequately and timely corrected. Kroksmark *et al.* has reported a group of patients with amyoplasia that are indistinguishable in the newborn period and respond dramatically to physical therapy.<sup>12</sup> All of these series suggest an optimistic childhood, with peak function occurring by the age of 10 years and some decline in ambulation into the adult years. Despite these reports, many children with amyoplasia will not have ambulatory potential. Assessment for this potential can be difficult, with decisions of the extent and timing of surgery hanging in balance. Serial evaluations involving physical therapy are important.

This study used a retrospective design. Therefore, it had limited control over the nature and quality of data collected. We found residual knee flexion contracture made patient nonambulatory. Future multicenter studies by exploring the effects and involvement of residual deformity which affect ambulation in this population are recommended. Longer followup periods should be included to verify the extent to which ambulatory gains are maintained.

Knee involvement is commonly seen among children with arthrogyriposis, with flexion contracture of the knee being the most frequent knee deformity. Knee flexion contracture in the pediatric population is particularly debilitating as it adversely affects ambulation. Treatment for knee flexion contracture requires numerous orthopedic procedures and extensive followup period. According to most authors, knee flexion contracture is more frequent and more difficult to treat with conservative methods than the knee extension contracture.<sup>13,14</sup> In the literature consensus is to perform early serial casting and physiotherapy, which sometimes is sufficient to correct contracture in flexion or extension. In cases of failure of conservative treatment surgery is recommended and includes procedures like by posterior capsulotomy in addition to soft-tissue release (including hamstring tenotomy). Del-Bello and Watts recommended performing a distal femoral extension osteotomy for flexion contracture as a safe procedure even for severe contractures.<sup>15</sup> Thomas *et al.* used femoral osteotomies in young patients because the walking improvement is large and quick (32 nonambulators became community or household ambulators in their series).<sup>13</sup> Brunner *et al.* used the Ilizarov technique as an efficient tool for knee and foot deformity corrections; however, he noted a high recurrence rate in children younger than 10 years of age.<sup>16</sup> Thomas *et al.* and Hoffer *et al.* noted the importance of having a flexion contracture less than 20 degrees in order to walk independently.<sup>9,13</sup> Our results support this conclusion. Murray and Fixsen presented functional

results of 22 patients with amyoplasia followed up over an average of 7 years. Patients in this series were treated by initial physiotherapy and splintage, as well as repeated posterior release and bony procedures when necessary. They reported 14 community walkers in 22 patients.<sup>14</sup>

Herzenberg *et al.* reported 10 patients with knee flexion contractures of various different etiologies, including 2 patients with arthrogryposis, treated with Ilizarov external fixator. Both became community ambulators at the latest followup.<sup>17</sup>

A study by van Bosse *et al.*<sup>18</sup> reported a high rate of improvement in ambulation in all seven children with arthrogryposis with knee flexion contracture after Ilizarov correction at latest followup, including five who were initially nonambulatory. A recent report by Fassier *et al.* indicated that among 11 children with arthrogryposis, 7 were independent ambulators at skeletal maturity.<sup>19</sup> However, these children had normal knee mobility or a flexion contracture of less than 15° and most had mild involvement of the hips, spine, or upper extremities. The two nonwalkers in their series had severe scoliosis treated by spinal fusion. We agree with these researchers that early aggressive management of children with severe arthrogryposis is justified and recommended.

In our series hip flexion contracture is a significant predictor of amputation. It may affect ambulatory status if a residual deformity has been left. We observed that community ambulators presented with less than 30 degrees of hip flexion contracture. Hoffer *et al.* also noted that less than 30° of hip flexion contracture in their independently walking patients.<sup>9</sup>

Staheli *et al.*<sup>4,20</sup> and Hoffer *et al.*<sup>9</sup> have categorized children with ambulatory potential as having grade 4 or greater hip extensors; hip flexion contracture less than 20°; grade 4 quadriceps function; grade 3 quadriceps function with knee flexion contracture less than 20°; and good strength and sitting balance, with shoulder depressor function and only mild upper extremity abnormalities but many studies show progressive muscle activity improvement after the recovery of a passive range of motion in a useful arc. Many studies showed that independent ambulators were able to perform an increase in strength during growth. The difficulty remaining in identifying prognosis factors is, as with the case of any rare diseases, the small number of patients available to generate data. In our series, poor prognostic factors for independent or community walkers were residual knee flexion contracture more than 30°.

Hip flexion contracture is less likely to occur in isolation, but if greater than 30°, it may affect ambulation, and if greater than 45°, it will generally require release. Often, the combination of hip flexion, external rotation, and abduction contractures

are present.<sup>21</sup> In these cases, the hip abductors may function as extensors and if the hip is left in external rotation, the hip can often be adducted to neutral and extend to less than 30° of contracture without soft tissue release while preserving good hip extensor strength for standing. A distal femoral osteotomy is necessary to bring the knee and foot into alignment for ambulation. Reduction of hip dislocation is recommended for unilateral dislocation in all reports.<sup>20,21</sup>

Long term ambulatory status at skeletal maturity is not correlated with the severity of condition at birth.<sup>19</sup> Therefore, we believe that early aggressive management of children with severe arthrogryposis is warranted and justified. This includes physiotherapy, occupational therapy and multiple surgeries. The ambulatory status in children with arthrogryposis is variable and our findings seem to indicate that although knee flexion contractures have an impact on ambulation, hip flexion contracture may also influence ambulation. Our findings emphasize that residual knee flexion contracture affects the ambulatory status of children with arthrogryposis. Surgical interventions for knee flexion contractures in this population is warranted and support a positive prognosis.

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